Principles of Inheritance and Variation

INTRODUCTION OF GENETICS :

Genetics term was given by **W. Bateson.**

Genetics: Collective study of heredity & Variations.

Heredity: Transmission of genetic characters from parents to offspring.

Variation: Individuals of same species have some differences, these are called variation.

HISTORY OF RESEARCHES IN GENETICS :

G.J. Mendel: Father of Genetics, born in Austria.

W. Bateson: Father of Modern Genetics.

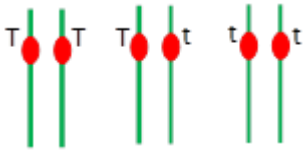
Morgan: Father of Experimental genetics, He performed experiments on Drosophila & proposed various concepts, like Linkage, Sex linkage, Crossing over, Criss - cross inheritance, Linkage map on Drosophila.

A. Garrod: Father of human genetics & Biochemical genetics. Garrod discovered first human Metabolic genetic disorder, which is called alkaptonuria (black urine disease).

SOME GENETICAL TERMS :

1. **FACTORS** :- Unit of heredity which is responsible for inheritance and appearance of characters. These factors were referred as genes by **Johannsen(1909)**. Mendel used term "**element**" or "factor. Morgan first used symbol to represent the factor. Dominant factors are represented by capital letter while recessive factor by small letter.

2. **ALLELE** :- Alternative forms of a gene which are located on same position [loci] on the homologous chromosome are called Allele. Term allele was coined by Bateson.



3. **HOMOZYGOUS** :- A zygote is formed by fusion of two gametes having identicle factors is called homozygote and organism developed from this zygote is called homozygous. Ex. TT, RR, tt

4. **HETEROZYGOUS** :- A zygote is formed by fusion of two different types of gamete carrying different factors is called heterozygote (**Tt, Rr**) and individual developed from such zygote is called heterozygous. The term homozygous and heterozygous are coined by Bateson.

5. **HEMIZYGOUS** :- If any diploid individual contains only one gene of a pair then individual is said to be Hemizygous. Male individual is always Hemizygous for sex linked gene.

6. **PHENOTYPE** :- It is the external and morphological appearance of an organism for a particular character.

7. **GENOTYPE** :- The genetic constitution or genetic make-up of an organism for a particular character. Genotype & phenotype terms were coined by Johannsen.

8. **PHENOCOPY** :- If different genotypes are placed in different environmental conditions then they produce same phenotype. Then these are said to be Phenocopy.

9. **HYBRID VIGOUR/HETEROSIS** :-Superiority of offsprings over it's parents is called as **Hybrid vigour** & it develops due to Heterozygosity. Hybrid vigour can be maintained for long time in vegetative propagated crops. Hybrid vigour can be lost by inbreeding (selfing) because inbreeding induces the Homozygosity in off springs. Loss of Hybrid vigour due to inbreeding, is called as **inbreeding depression**.

Humans knew from as early as 8000-1000 B.C. that one of the causes of variation was hidden in sexual reproduction. They exploited the variations that were naturally present in the wild populations of plants and animals to selectively breed and select for organisms that possessed desirable characters. For example, through artificial selection and domestication from ancestral wild cows, we have well-known Indian breeds, e.g., Sahiwal cows in Punjab. We must, however, recognise that though our ancestors knew about the inheritance of characters and variation, they had very little idea about the scientific basis of these phenomena.

Question-1: When a plant have two different alleles of a gene it is called :-

(1) Homozygous
(2) Dioecious
(3) Heterozygous
(4) Monoecious

Answer: (3) Heterozygous

Question-2: Term "Genetics" was given by :-

(1) W. Bateson.
(2) G.J. Mendel
(3) Morgan
(4) A. Garrod

Answer: (1) W. Bateson.

Question-3: Two allelic genes are located on :-

(1) The same chromosome
(2) Two homologous chromosomes
(3) Two-non-homologous chromosomes
(4) Any two chromosomes

Answer: (2) Two homologous chromosomes

Question-4: An individual with two identical members of a pair of genetic factors is called :-

(1) Hemizygous
(2) Heterozygous
(3) Nonhomologous
(4) Homozygous

Answer: (1) Homozygous

MENDELISM :

Gregor Johann Mendel (1822 - 1884) :- Mendel was born on July 22,1822 at **Heinzen Dorf** in Austria at Silesia village. Mendel worked in Augustinian Monastery as monk at **Brunn city, Austria.**

- In **1856-57,** he started his historical experiments of heredity on **pea (*Pisum sativum*)** plant. His experimental work continued on pea plant till **1863** (19th century).
- The results of his experiments were published in **1865**.
- A paper of Mendel by the name of "**Experiment in plant Hybridization**" was published in this journal. Mendel was unable to get any popularity. No one understood of him. He died in 1884 without getting any credit of his work.

- After 16 years of Mendel's death in 1900, Mendel's postulates were rediscovered. Rediscovery by three scientists independently.

- **1. Carl Correns** - (Germany) - (Experiment on Maize)
- **2.Hugo de Vries** (Holland) (Experiment on Evening Primrose) He republished Mendel's results in 1901 in Flora magazine
- **3.Erich von Tschermak** - (Austria) (Experiment on different flowering plants).

The credit of rediscovery of Mendelism goes to three scientists.

Carl Correns converted two postulates of Mendel into :–

LAW OF HEREDITY/INHERITANCE/MENDELISM :

Ist Law - Law of segregation.

IInd Law - Law of independent assortment.

Experiments performed by Mendel on genetics and description of mechanisms of heredity processes and formulation of principles are known as Mendelism. Mendel postulated various experimental laws in relation of genetics.

MENDEL STUDIED 7 CHARACTERS OR 7 PAIRS OF CONTRASTING TRAITS IN PEA (*PISUM SATIVUM*)

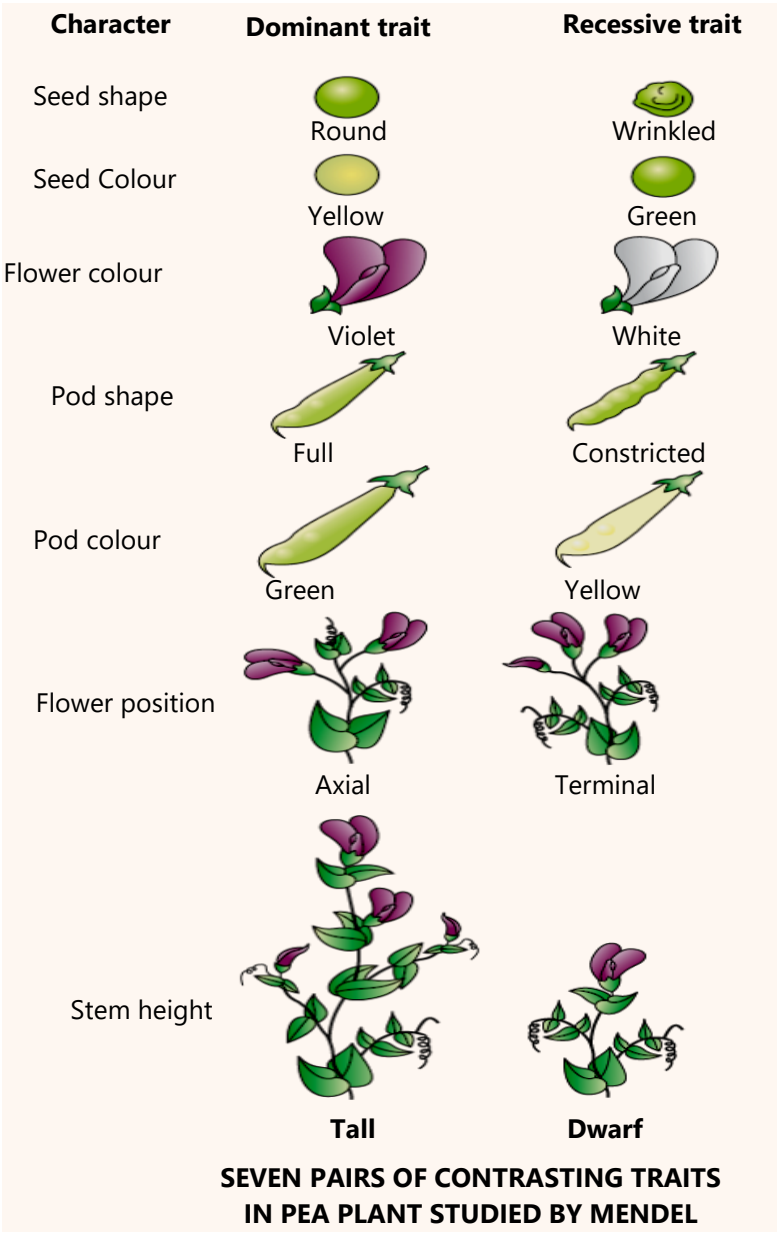
S.No.	Character	Ch. No.	Dominant	Recessive	Ratio
1.	Length of plant (Stem height)	**4th**	787 (tall)	277 (dwarf)	2.84:1
2.	Flower position	**4th**	651 (axial)	207 (terminal)	3.14:1
3.	Shape of pod	**4th**	882 (inflated)	299 (constricted)	2.94:1
4.	Pod colour	**5th**	4228 (green)	152 (yellow)	2.82:1
5.	Seed shape	**7th**	5.474(round)	1.850 (wrinkled)	2.96:1
6.	Seed colour	**1st**	6.022(yellow)	2.001 (green)	3.01:1
7.	Flower colour	**1st**	705 (violet)	224 (white)	3.15:1
Average of all traits studied					**2.98(=3:1)**

SPECIAL POINT:

S. Blixt concluded that the genes studied by Mendel are located on four different pairs of chromosomes. These are chromosome 1st, 4th, 5th, 7th Two of the genes are on chromosome 1st and three are on chromosome 4th. These genes are located far apart on the chromosome except genes controlling plant height and pod shape. In Pea plant sead coat colour and Flower colour are regulated by same gene. Gene which controls more than one character is called as **pleiotropic gene.**

Mendel obtained wrinkled seeds due to absence of **Starch Branching enzyme (SBE)**

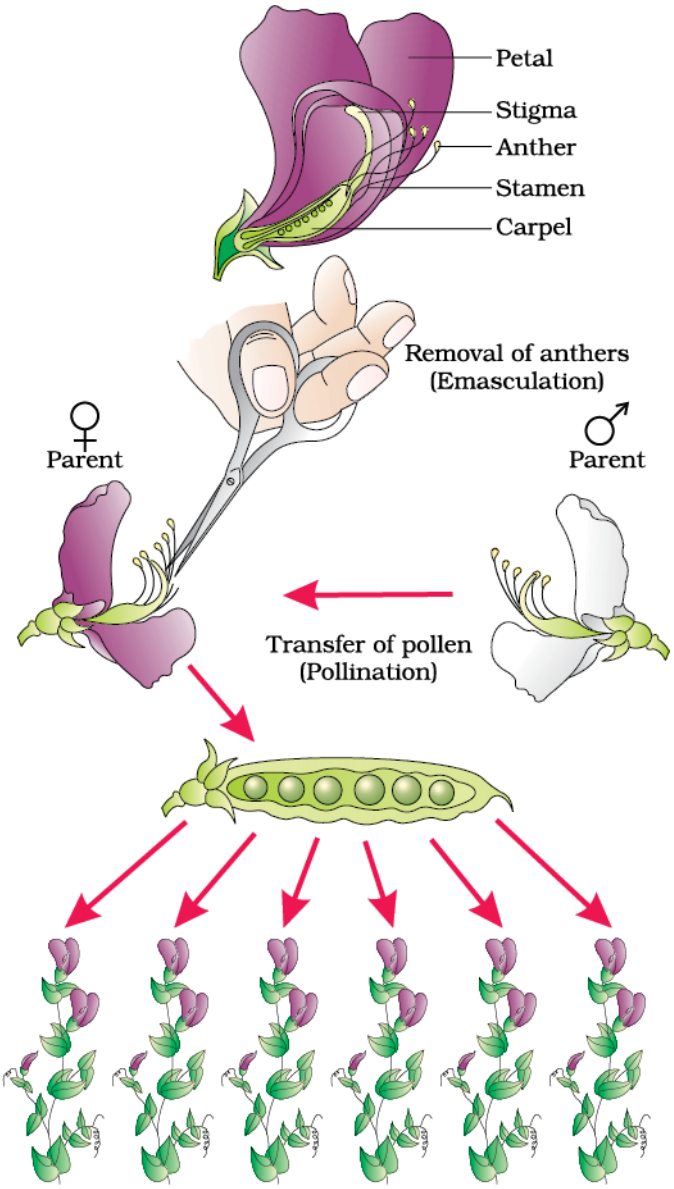
In Wrinkled seed free sugar is more in place of starch.



SEVEN PAIRS OF CONTRASTING TRAITS IN PEA PLANT STUDIED BY MENDEL

TECHNIQUE OF MENDAL :

- Mendel conducted such artificial pollination/crosspollination experiments using several true-breeding pea lines. A true breeding line is one that, having undergone continuous self-pollination, shows the stable trait inheritance and expression for several generations. Mendel selected 14 true-breeding pea plant varieties, as pairs which were similar except for one character with contrasting traits.



- He developed a technique **Emasculation and Bagging** for hybridization in plants. Flowers of pea plant are bisexual. In this method one considered as male and another as female.
- **Emasculation:** Removal of anther from a bisexual flower in immature stage is called Emasculation. Emasculation is done to prevent self-pollination.
- **Bagging**: Emasculated flowers are covered by bags, this is called bagging. Bagging is only used to prevent undesirable cross pollination. Mature pollen grains are collected from male plants and spread over emasculated flower.

  Seeds are formed in the female flower after pollination.

  The plants that are obtained from these seeds are called First Filial generation or F1 generation according to Mendel.

  The plants of F1 generation are self-pollinated & F2 generation is produced.

Mendel published his work on inheritance of characters in 1865 but for several reasons, it remained unrecognised till 1900.

- Firstly, communication was not easy (as it is now) in those days and his work could not be widely publicised.
- Secondly, his concept of genes (or factors, in Mendel's words) as stable and discrete units that controlled the expression of traits and, of the pair of alleles which did not 'blend' with each other, was not accepted by his contemporaries as an explanation for the apparently continuous variation seen in nature.
- Thirdly, Mendel's approach of using mathematics to explain biological phenomenon was totally new and unacceptable to many of the biologists of his time.
- Finally, though Mendel's work suggested that factors (genes)were discrete units, he could not provide any physical proof for the existence of factors or say what they were made of.

REASONS FOR MENDEL'S SUCCESS :

1. Mendel studied the inheritance of one or two characters at a time unlike his predecessors who had considered many characters at a time. (Kolreuter-Tobacco plant, John Goss & Knight -Pea plant).

2. **Selection of Material:**

Selection of garden Pea plant is suitable for studies, which have the following advantages :

- **(i)** Pea plant is annual plant with short life cycle of 2-3 months so large no. of off springs can be analysed within a short period of time.
- **(ii)** It has many contrasting traits.
- **(iii)** Natural self pollination is present in pea plant.
- **(iv)** Cross pollination can be performed in it artificially so hybridization can be made possible.
- **(v)** Pea plant is easy to cultivate.
- **(vi)** Pea seeds are large. In addition to pea, Mendel worked on rajma and hawk weed.

3. Mendel quantitatively analyse the inheritance of qualitative characters.

4. He maintained the statistical records of all the experiments.

5. His experiments had a large sampling size.

Gregor Mendel, conducted hybridisation experiments on garden peas for seven years (1856-1863) and proposed the laws of inheritance in living organisms.
During Mendel's investigations into inheritance patterns it was for the first time that statistical analysis and mathematical logic were applied to problems in biology.
His experiments had a large sampling size, which gave greater credibility to the data that he collected.
Mendel investigated characters in the garden pea plant that were manifested as two opposing traits, e.g., tall or dwarf plants, yellow or green seeds.
Mendel conducted such artificial pollination/cross pollination experiments using several true-breeding pea lines.
A truebreeding line is one that, having undergone continuous self-pollination, shows the stable trait inheritance and expression for several generations.
Mendel selected 14 true-breeding pea plant varieties.
As pairs which were similar except for one character with contrasting traits.

Question-1: During breeding the removal of anthers from a flower is called :-

(1) Anthesis
(2) Pollination
(3) Emasculation
(4) Vasectomy

Answer: (3) Emasculation

Question-2: How many characters of pea pod were chosen by Mendel?

(1) 7
(2) 5
(3) 4
(4) 2

Answer: (4) 2

Question-3: When flowers are unisexual then emasculation is done in?

(1) Female
(2) Male
(3) 1 & 2 both
(4) None of these

Answer: (4) None of these

Question-4: Mendel published his work on inheritance of characters in :-

(1) 1900
(2) 1857
(3) 1902
(4) 1865

Answer: (4) 1865

Question-5: Main reason of Mendel's success was that :-

(1) Statistical analysis
(2) Study of one or two character at a time
(3) Pedigree records
(4) Selection of pea plant

Answer: (2) study of one or two character at a time

Question-6: Pea plant is suitable for studies because :-

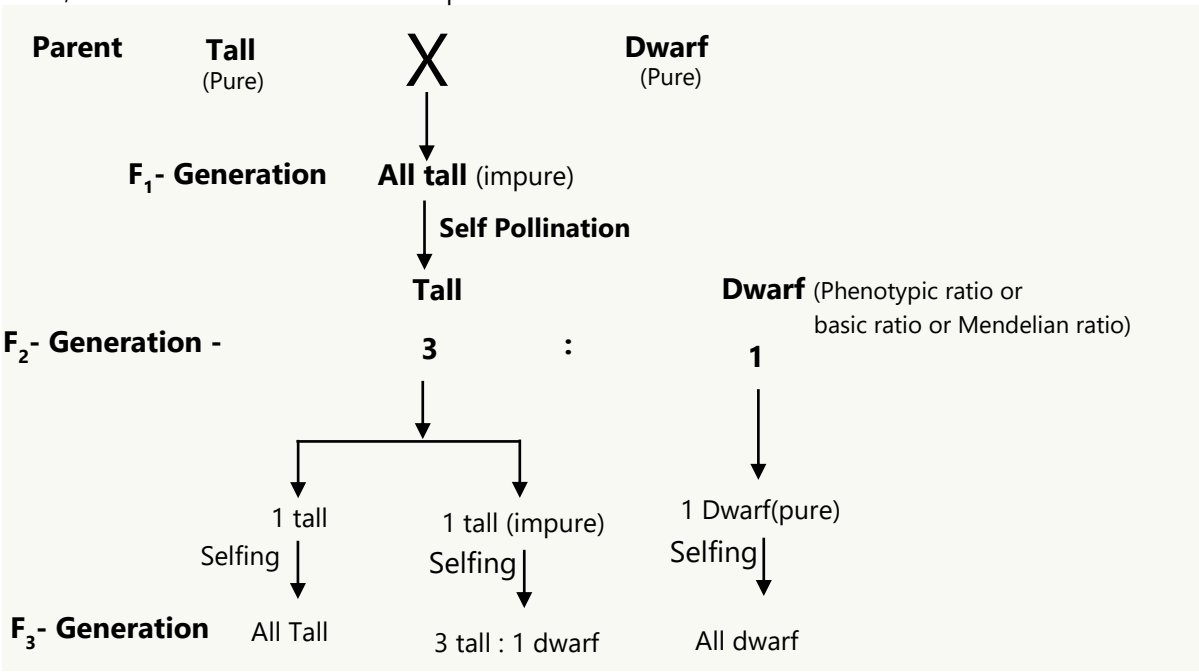
(1) It has many contrasting traits.
(2) Natural self-pollination is present in pea plant.
(3) Pea plant is easy to cultivate.
(4) All of above

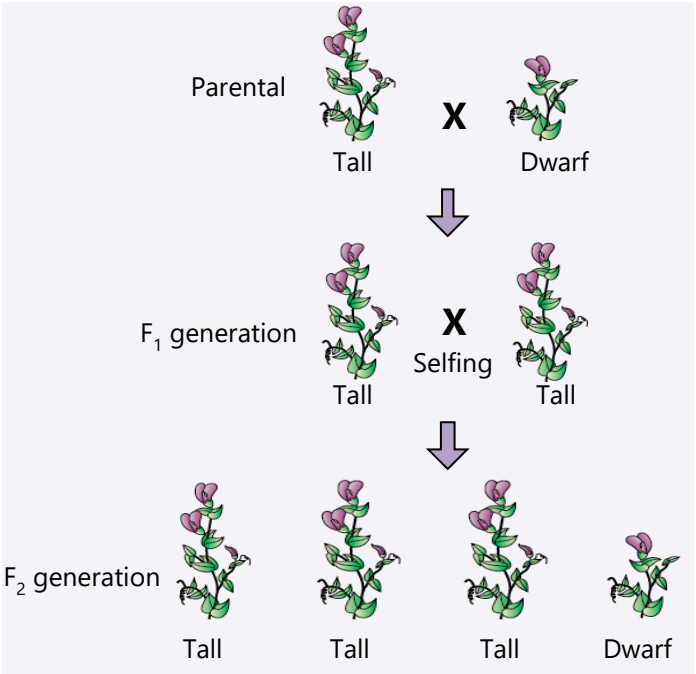
Answer: (4) All of above

INHERITANCE OF ONE GENE (MONOHYBRID CROSS) :

INTRODUCTION :

When we consider the inheritance of one character at a time in a cross this is called monohybrid cross. First of all, Mendel selected tall and dwarf plants.





CHECKER BOARD METHOD :

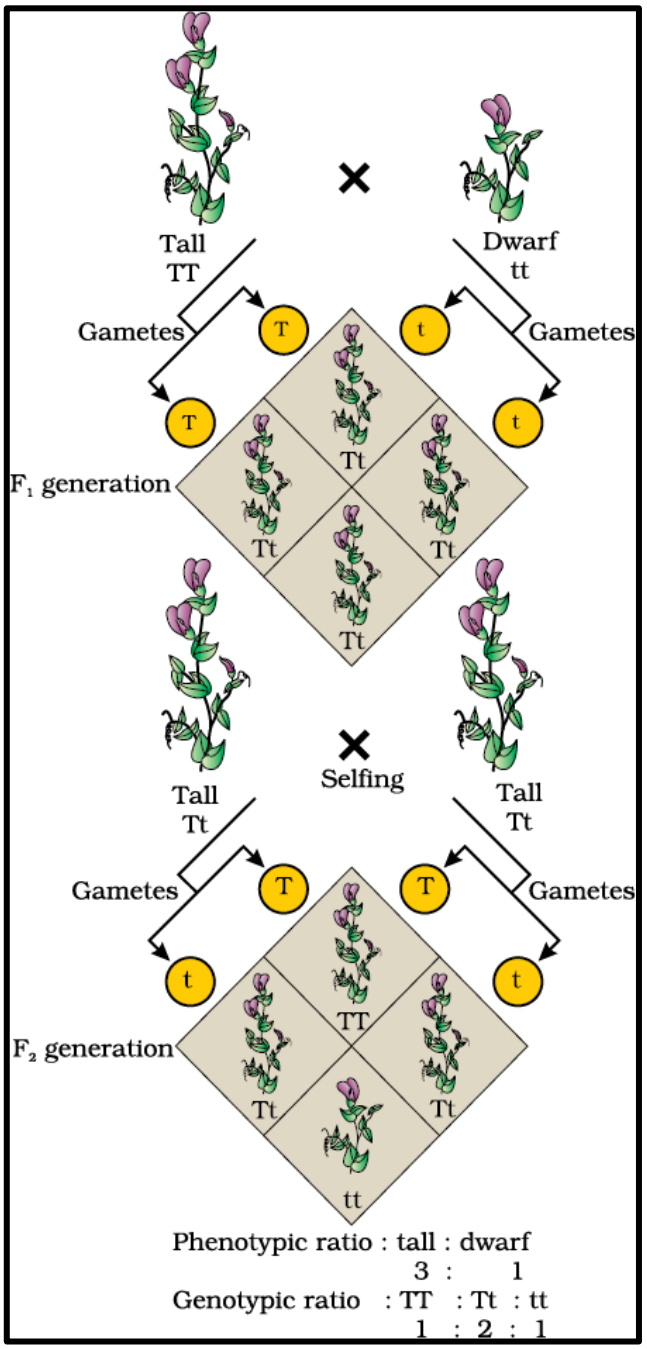
First time, it was used by **Reginald. C.Punnett (1875 - 1967)** The representation of generations to analyse in the form of symbols of squares. Male gametes lie horizontally and female gametes lie vertically.

PHENOTYPIC RATIO : tall : dwarf

1 : 3

GENOTYPIC RATIO : TT : Tt : tt

1 : 2 : 1



Conclusions (results) of Monohybrid Cross

I[st] Conclusion (Postulate of paired factors)

II[nd] Conclusion (Postulate of Dominance)

III[rd] Conclusion (Law of segregation)

LAW OF DOMINANCE :

CONCLUSIONS (RESULTS) OF MONOHYBRID CROSS :

I[st] CONCLUSION (POSTULATE OF PAIRED FACTORS) :

According to Mendel :-

1. Characters are controlled by discrete units called factors.
2. Factors occur in pairs.

IInd CONCLUSION (POSTULATE OF DOMINANCE) :

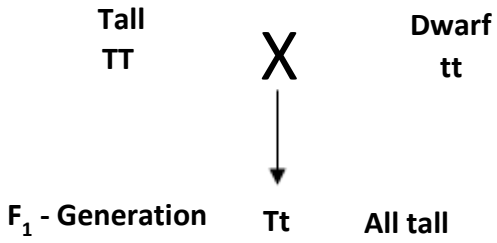
This conclusion is based on F1 - generation. When two different unit factors are present in single individual, only one-unit factor is able to express itself and known as dominant unit factor. Another unit factor fails to express is the recessive factor. In the presence of dominant unit factor recessive unit factor cannot express and it is known as conclusion of dominance.

Law of dominance = Ist Conclusion + IInd Conclusion.

There are two exceptions of law of dominance.

[A] Incomplete dominance

[B] Co-dominance.



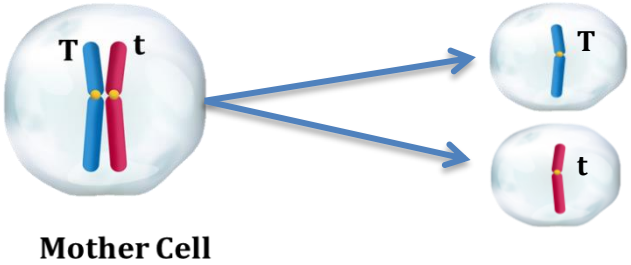
Law of Dominance

(i) Characters are controlled by discrete units called factors.
(ii) Factors occur in pairs.
(iii) In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).
The law of dominance is used to explain the expression of only one of the parental characters in a monohybrid cross in the F_1 and the expression of both in the F_2. It also explains the proportion of 3 : 1 obtained at the F_2.

IIIrd CONCLUSION (LAW OF SEGREGATION) :

This law is based on the fact that the alleles do not show any blending and that both the characters are recovered as such in the F_2 generation. During gamete formation; the unit factors of a pair segregate randomly and transfer inside different gamete.

Each gamete receives only one factor of a pair; so, gametes are pure for a particular trait. It is known as conclusion of purity of gametes or segregation.

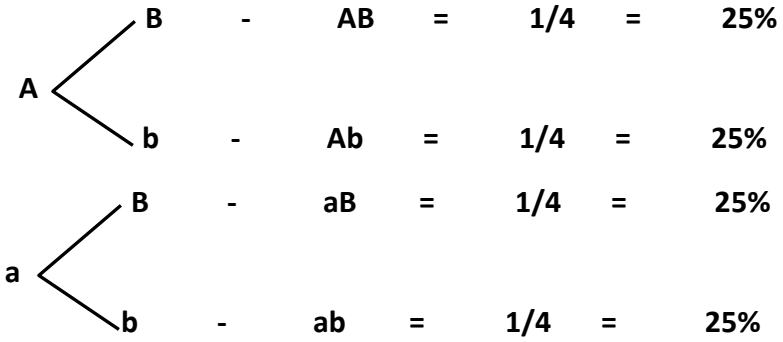


There is no exception of Law of segregation. The segregation is essential during the meiotic division in all sexually reproducing organisms. (Nondisjunction may be exception of this law).

FORK LINE METHOD FOR GAMETES FORMATION :

To find out the composition of factors inside the gamete, we use fork line method.

AaBb = 4 types of gamete



Type of gamete /phenotypic category = 2^n
Type of genotype = 3^n
Number of zygote produced by selfing of a genotype = 4^n
n = Number of hybrid character or heterozygous pair.

Law of Segregation

This law is based on the fact that the alleles do not show any blending and that both the characters are recovered as such in the F_2 generation though one of these is not seen at the F_1 stage. Though the parents contain two alleles during gamete formation, the factors or alleles of a pair segregate from each other such that a gamete receives only one of the two factors. Of course, a homozygous parent produces all gametes that are similar while a heterozygous one produces two kinds of gametes each having one allele with equal proportion.

Question-1: In monohybrid cross what is the ratio of homozygous dominant and homozygous recessive individuals in F_2– generation :-

(1) 1 : 2 : 1
(2) 2 : 1 / 1 : 2
(3) 3 : 1 / 1 : 3
(4) 1 : 1

Answer: (4) 1 : 1

Question-2: If 120 Plants are produced on crossing pure red and pure white flowered pea plants, than the ratio of off springs will be : - (Red is dominant)

(1) 90 Red : 30 White
(2) 30 Red : 90 White
(3) 60 Red : 60 White
(4) All Red

Answer: (4) All Red

Question-3: **What will be genotype of parental garden pea plant if they produce 44 tall and 15 dwarf off spring's?**

(1) TT x TT

(2) Tt x Tt

(3) TT x tt

(4) Tt x tt

Answer: **(2) Tt x Tt**

Question-4: **In garden pea plant, a pure violet flower plant is crossed with white flower plant. In F_2 generation 1000 offspring are obtained. How many of them have :-**

(a) Pure flowered plant

(b) Impure violet flowered plant

(c) White flowered plant

Answer: **(a) 500, (b) 500, (c) 250**

Question-5: **The allele which is unable to express its effect in the presence of another is called :-**

(1) Co-dominant

(2) Supplementary

(3) Complementary

(4) Recessive

Answer: **(4) Recessive**

Question-6: **A gene said to be dominant if :-**

(1) It expresses it's effect only in homozygous stage.

(2) It expressed only in heterozygous condition

(3) It expressed both in homozygous and heterozygous condition.

(4) It never expressed in any condition

Answer: **(3) It expressed both in homozygous and heterozygous condition.**

Question-7: **Which one of the following traits of garden pea studied by Mendel, was a recessive feature :-**

(1) Axial flower position

(2) Green seed colour

(3) Green pod colour

(4) Round seed shape

Answer: **(2) green seed colour**

Question-8: **Genes do not occur in pairs in :-**

(1) Zygote

(2) Somatic cell

(3) Embryo

(4) Gametes

Answer: **(4) Gametes**

Question-9: Segregation of genes take place during :-

(1) Metaphase

(2) Anaphase

(3) Prophase

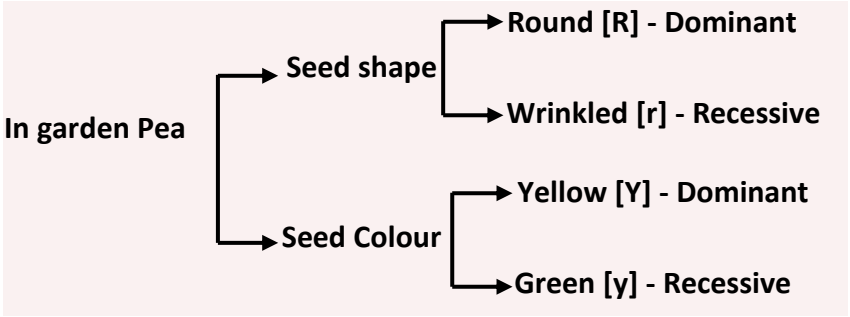
(4) Embryo formation

Answer: (2) Anaphase

INHERITANCE OF TWO GENES (DIHYBRID CROSS)

A cross in which there is study of inheritance of two pairs of contrasting traits. Mendel wanted to observe the effect of one pair of heterozygous on other pair.

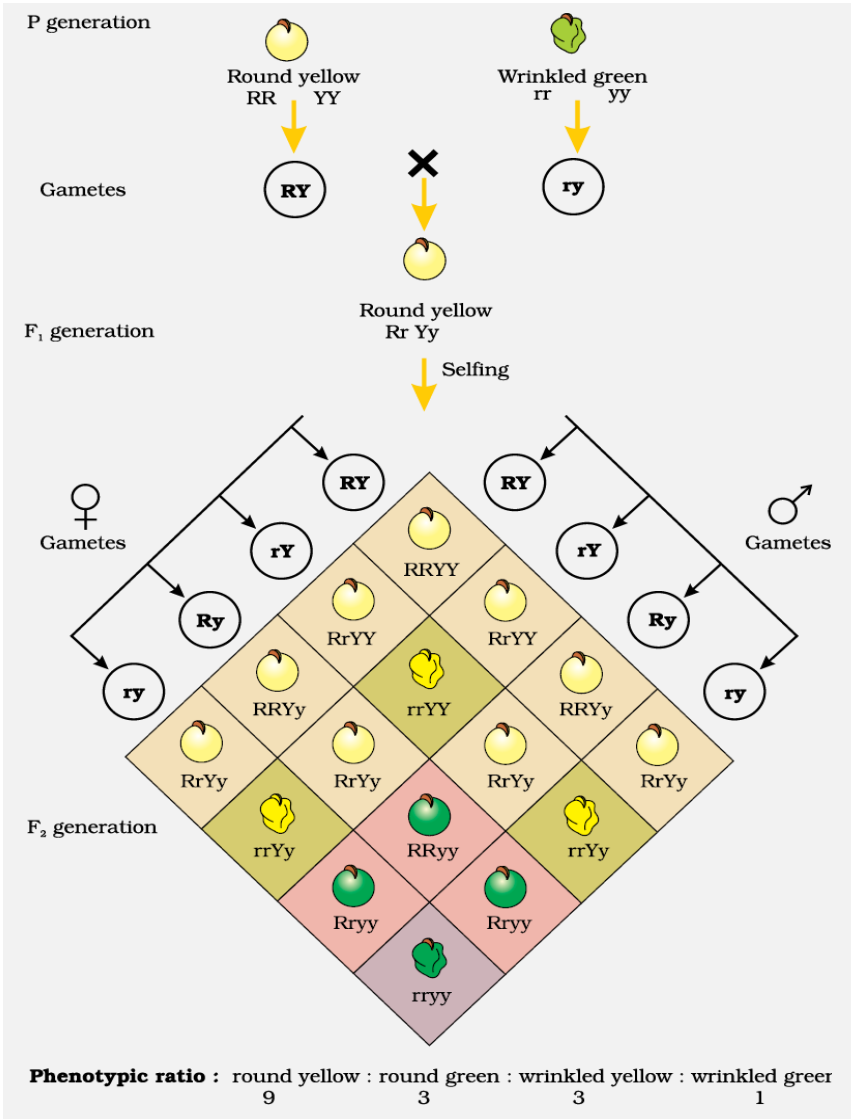
MENDEL SELECTED :-



Mendel crossed, pure yellow and round seeded plants with green and wrinkled seeded plants.

All the plants **in F_1 –generation had yellow and round seeds.**

When F_1 plants were self pollinated to produce four kinds of plants in F_2 generation such as **yellow round, yellow–wrinkled, green round and green wrinkled, there were in the ratio of 9 : 3 : 3 : 1.** This ratio is known as dihybrid ratio.



PHENOTYPIC RATIO :

Round yellow : round green : wrinkled yellow : wrinkled green

9 3 3 1

Number of phenotype = 4

GENOTYPIC RATIO :

RRYY : RrYY : RRYy : RrYy : rrYY : rrYy : RRyy : Rryy : rryy

1 2 2 4 1 2 1 2 1

Number of genotype = 9

Parental combination : New combination

9[Round yellow] + 1 [Green wrinkled] : 3[Round green] + 3 [Wrinkled yellow]

10 : 6

Note : Mendel obtained new combinations in F_2 generation due to independent assortment.

Mendel also worked with and crossed pea plants that differed in two characters, as is seen in the cross between a pea plant that has seeds with yellow colour and round shape and one that had seeds of green colour and wrinkled shape. Mendel found that the seeds resulting from the crossing of the parents, had yellow coloured and round shaped seeds. *Here can you tell which of the characters in the pairs yellow/green colour and round/wrinkled shape was dominant?*

Law of Independent Assortment :

The law states that "When two pairs of trait [2 different characters] are combined in a hybrid, segregation of one pair of character is independent of the other pair of character. "This is known as **Conclusion of Independent Assortment**. It is based on F_2 - generation of dihybrid cross.

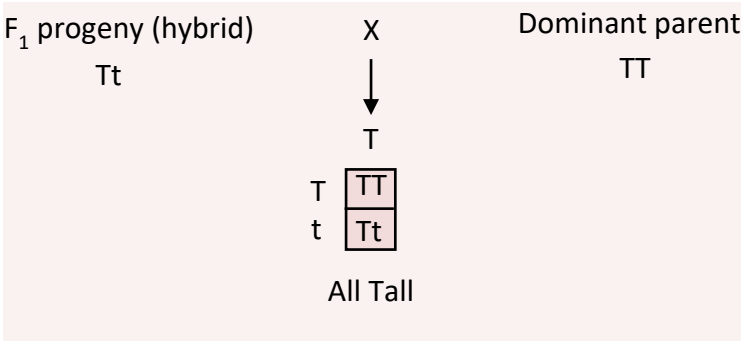
Exception : **Linkage.**

In the dihybrid cross, the phenotypes round, yellow; wrinkled, yellow; round, green and wrinkled, green appeared in the ratio 9 : 3 : 3 : 1. Such a ratio was observed for several pairs of characters that Mendel studied. The ratio of 9:3:3:1 can be derived as a combination series of 3 yellow : 1 green, with 3 round : 1 wrinkled. This derivation can be written as follows: (3 Round : 1 Wrinkled) (3 Yellow : 1 Green) = 9 Round, Yellow : 3 Wrinkled, Yellow: 3 Round, Green : 1 Wrinkled, Green

BACK CROSS

A back cross is a cross in which F_1 individuals are crossed with any of their parents.

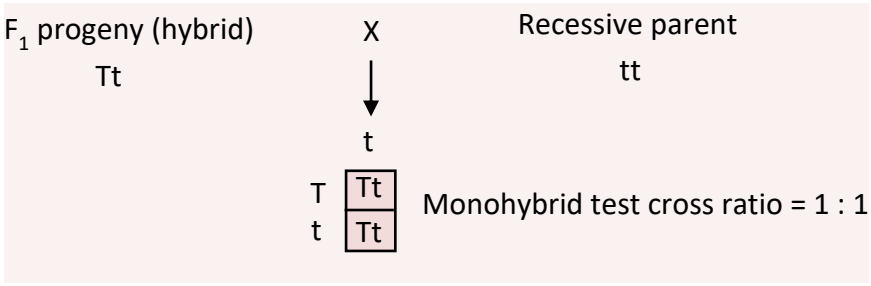
(1) OUT CROSS : When F_1 (heterozygous) individual is crossed with dominant parent then it is termed out cross. The generations obtained from this cross, all possess dominant character. So, the any analysis cannot possible in F_1 generation.



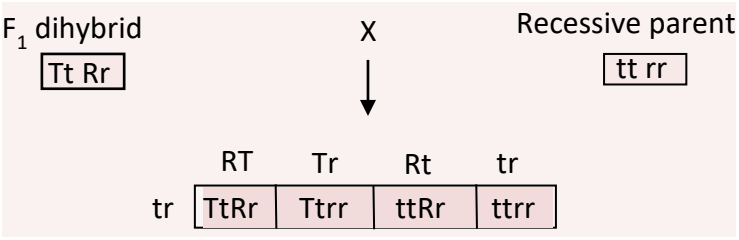
(2) TEST CROSS: When F1 progeny (heterozygous) is crossed with recessive parent then it is called test cross.

a. Monohybrid Test Cross: - The progeny obtained from the monohybrid test cross are in equal proportion, means 50% is dominant phenotypes and 50% is recessive phenotypes.

It can be represented in symbolic forms as follows.

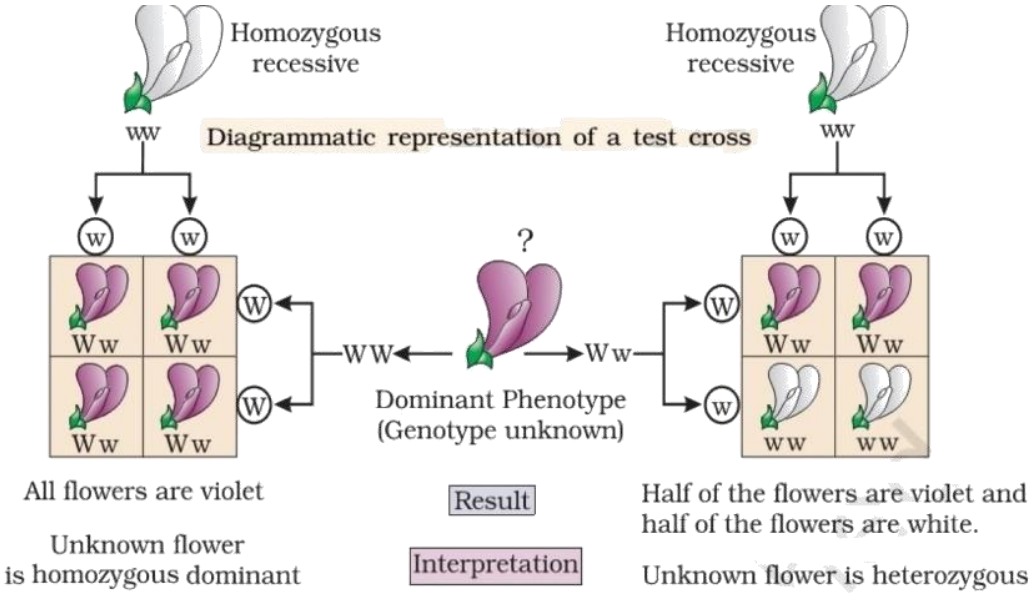


b. Dihybrid Test Cross: - The progeny is obtained from dihybrid test cross are four types and each of them is 25%.



The ratio of Dihybrid test cross = 1 : 1 : 1 : 1

Conclusion: - In test cross phenotypes and genotypes ratio are same. Test cross helps to find out the genotype of dominant individual.



To determine the genotype of a tall plant at F_2, Mendel crossed the tall plant from F_2 with a dwarf plant. This he called a test cross. In a typical test cross an organism (pea plants here) showing a dominant phenotype (and whose genotype is to be determined) is crossed with the recessive parent instead of self-crossing. The progenies of such a cross can easily be analysed to predict the genotype of the test organism.

Question-1: A plant with genotype DdEe is crossed with other plant having genotype Ddee. Find out probability of offspring's which have following genotype :-

Ddee, DDEe, DDEE :-

(1) 25%,12.5%, 0%

(2) 12.5%, 0%, 25%

(3) 12.5%, 25%, 0%

(4) 0%, 12.5%, 25%

Answer: (1) 25%,12.5%, 0%

Question-2: A plant with genotype RRHhGg is crossed with other plant having genotype rrhhGg and produce 800 pffsprings. Find out number of offspring's which have following genotype :-

RrhhGG, RRHhGg, RrHhGg

(1) 100, 0, 200

(2) 100, 100, 0

(3) 200, 0, 200

(4) 200, 200, 0

Answer: (1) 100, 0, 200

Question-3: In a cross between AaBb and aaBb genotypic plant. The ratio of AaBB, AABb, aaBb, aabb in offspring's is :-

(1) 9 : 3 : 3 : 1

(2) 1 : 1 : 1 : 1

(3) 1 : 0 : 2 : 1

(4) 1 : 1 : 2 : 2

Answer: (3) 1 : 0 : 2 : 1

Question-4: When F_1 generation hybrid tall Tt is crossed with dwarf tt parent, it is a case of :-

(1) Dihybrid cross

(2) Test cross

(3) Crossing over

(4) Reciprocal cross

Answer: (2) Test cross

Question-5: Which genotype represents a true dihybrid condition :-

(1) tt rr

(2) Tt rr

(3) Tt Rr

(4) TT Rr

Answer: (3) TtRr

Question-6: Due to the cross between TTRr × ttrr the resultant progenies showed how many percent plants would be, tall, red flowered :-

(1) 50%

(2) 75%

(3) 25%

(4) 100%

Answer: (1) 50%

GENE INTERACTION :

INCOMPLETE DOMINANCE AND CO-DOMINANCE :

Gene interaction is of two types:

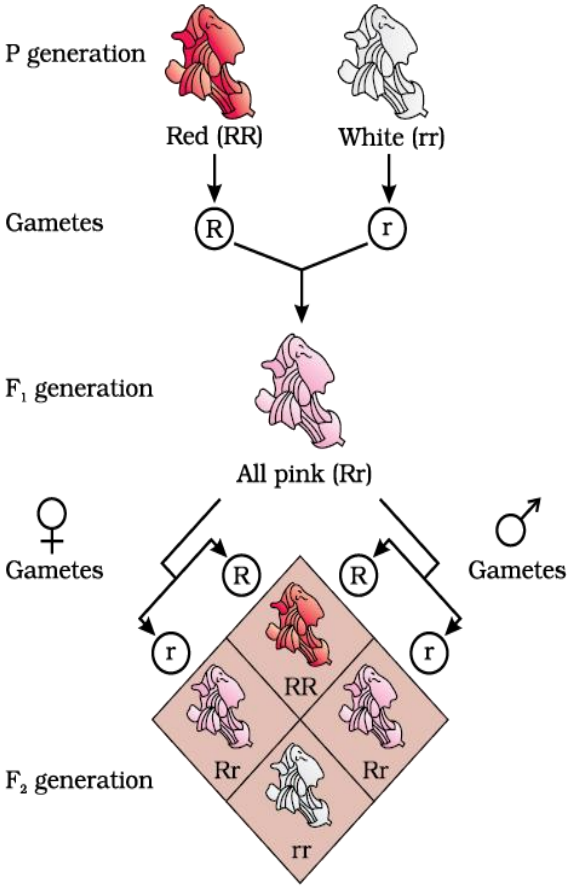
(i) Allelic interaction/Intragenic interaction

(ii) Non allelic interaction/Intergenic interaction

(i) **Allelic interaction/Intragenic interaction**: Allelic interaction takes place between allele of same gene which are present at same locus. Examples of Allelic interaction are as follows :-

1. **Incomplete dominance: -** According to Mendel's law of dominance, dominant character must be present in F1 generation. But in some organisms, F1 generation is different from the both parents. Both factors such as dominant and recessive are present in incomplete dominance but dominant factors is unable to express its character completely, resulting Intermediate type of generation is formed which is different from the both parents. Some examples are: -

a. **Flower colour in *Mirabilis jalapa* :-** Incomplete dominance was first discovered by **Correns in *Mirabilis jalapa*.** This plant is called as **'4 O' clock plant 'or' Gul-e-Bans'.** Three different types of plant are found in Mirabilis on the basis of flower colour, there are red, white and pink. When plants with red flowers is crossed with white flower, plants with pink flower are obtained in F1 generation. The reason of this is that the genes of red colour is incompletely dominant over the genes of white colour. When, F1 generation of pink flower is self-pollinated then the phenotypic ratio of F2 generation is **red, pink, white is 1:2:1** ratio in place of normal monohybrid cross **ratio 3:1. The ratio of phenotype and genotype of F2 generation in incomplete dominance is always same.**



Phenotypic ratio : red : pink : white
1 : 2 : 1

Genotypic ratio : RR : Rr : rr
1 : 2 : 1

b. **Flower colour in *Antirrhinum majus* :-** Incomplete dominance is also seen in flower colour of this plant. This plant is also known as 'Snapdragon ' or 'Dog flower'. Incomplete dominance is found in this plant which is the same as Mirabilis.

c. **Feather colour in Andalusian Fowls :-** Incomplete dominance is present for their feather colour. When a black colour fowl is crossed with a white colour fowl, the colour of F_1 generation is blue.

d. **Size of starch grain in pea plant :-** When a large size is crossed with small size, the size of starch grain in F_1 generation is intermediate.

When experiments on peas were repeated using other traits in other plants, it was found that sometimes the F_1 had a phenotype that did not resemble either of the two parents and was in between the two. The inheritance of flower colour in the dog flower (snapdragon or Antirrhinum sp.) is a good example to understand incomplete dominance. In a cross between true-breeding red-flowered (RR) and true breeding white-flowered plants (rr), the F_1 (Rr) was pink. When the F_1 was self-pollinated the F_2 resulted in the following ratio 1 (RR) Red: 2 (Rr) Pink: 1 (rr) White. Here the genotype ratios were exactly as we would expect in any mendelian monohybrid cross, but the phenotype ratios had changed from the 3:1 dominant : recessive ratio.

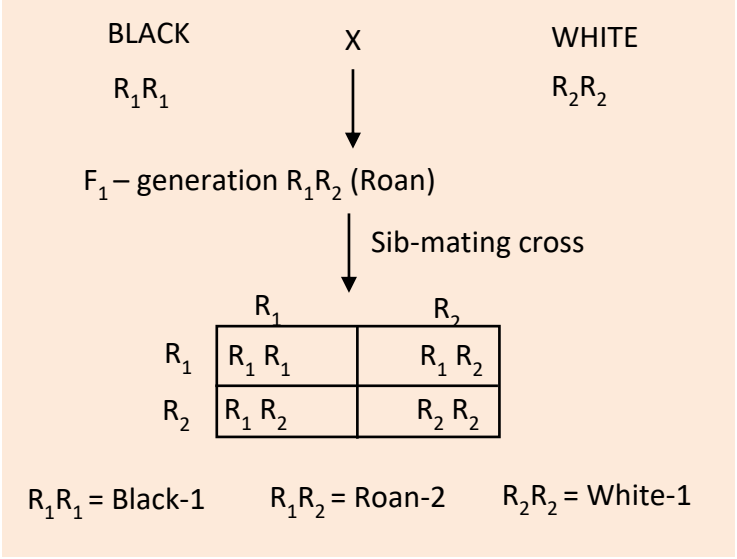
2. **Co-dominance:** - In this phenomenon, both the allele of a gene express for a particular character in F_1 progeny. There is no blending of characters, whereas both the characters expressed equally.

Examples :-

a. Co-dominance is seen in **cattle for coat colour**.

When a black parent is crossed with white parent, a roan colour F1 progeny is produced. When we obtain **F_2 generation from the F_1 generation, the ratio of black ; black-white (Roan) ; white cattle is obtained in 1 : 2 : 1**

Note :- F_2 generation is obtained in animals by **sib-mating cross**

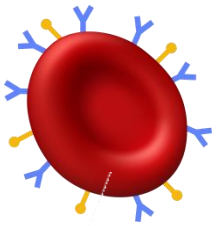


It is obvious by above analysis that the ratio of phenotype as well as genotype is 1:2:1 in co-dominance.

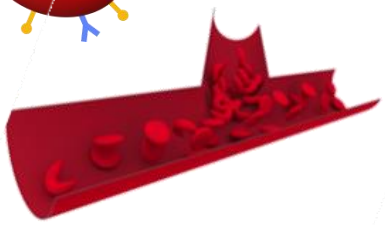
Sp. Note :- In incomplete dominance, characters are blended phenotypically, while in co-dominance, both the genes of a pair exhibit both the characters side by side and effect of both the character is independent from each other.

Other Examples of Co-dominance :

b. **AB blood group inheritance (I^AI^B)**



c. **Carrier of Sickle cell anaemia (Hb^A Hb^S)**



Till now we were discussing crosses where the F_1 resembled either of the two parents (dominance) or was in between (incomplete dominance). But, in the case of co-dominance the F_1 generation resembles both parents. A good example is different types of red blood cells that determine ABO blood grouping in human beings. ABO blood groups are controlled by the gene ***I***. The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene. The gene ***(I)*** has three alleles I^A, I^B and ***i***. The alleles I^A and I^B produce a slightly different form of the sugar while allele ***i*** does not produce any sugar.

Question-1: In *Mirabilis* red (RR) and white (rr) flower produces F_1 generation. Now F_1 generation is test crossed and produce 200 offspring's, how many of them are red, pink, and white flowered respectively :-

(1) 0, 100, 100

(2) 100, 0, 100

(3) 0, 100, 100

(4) 50, 50, 100

Answer: (1) 0, 100, 100

Question-2: With respect to size of starch grain in pea plant large sized starch garin plant is crossed with medium sized starch grain and produce 100 offsprings. Find out number of large, medium and small sized respectively?

(1) 0, 100, 0

(2) 0, 50, 50

(3) 50, 50, 0

(4) 100, 0, 0

Answer: (3) 50, 50, 0

Question-3: In a dihybrid cross, when one pair of alleles show incomplete dominance, genotypic ratio comes to

(1) 3 : 6 : 3 : 1 : 2 : 1

(2) 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

(3) 9 : 3 : 3 : 1

(4) 1 : 2 : 1

Answer: (2) 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

Question-4: **F_2 generation in a mendelian cross showed that both genotypic and phenotypic ratios are same as 1 : 2 : 1. It represents a case of :-**

(1) Monohybrid cross with complete dominance

(2) Monohybrid cross with incomplete dominance

(3) Dihybrid cross

(4) All of these

Answer: **(2)** Monohybrid cross with incomplete dominance

Question-5: **One di-hybrid cross is performed with two characters one character is complete dominant and another is incomplete dominant. Find out the phenotypic and genotypic ratio in F_2 generation.**

(1) Phenotypic Ratio :- 4 : 3 : 3 : 2 : 1 : 1, Genotypic Ratio :- 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

(2) Phenotypic Ratio :- 6 : 3 : 3 : 2 : 1 : 1, Genotypic Ratio :- 1 : 2 : 1 : 2 : 4 : 1 : 2 : 2 : 1

(3) Phenotypic Ratio :- 6 : 3 : 3 : 2 : 1 : 1, Genotypic Ratio :- 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

(4) Phenotypic Ratio :- 8 : 3 : 6 : 2 : 1 : 1, Genotypic Ratio :- 1 : 2 : 1 : 2 : 4 : 2 : 2 : 2 : 1

Answer: **(3)** Phenotypic Ratio :- 6 : 3 : 3 : 2 : 1 : 1, Genotypic Ratio :- 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

ALLELIC INTERACTION/ INTRAGENIC INTERACTION :

MULTIPLE ALLELISM AND PLEIOTROPY :

Multiple allele : More than 2 alternative forms of same gene are called as multiple allele. Multiple allele is formed due to mutation. Multiple allele are located on same locus of homologous chromosome. A diploid individual contains two alleles and gamete contains one allele for a character.

If n is the number of allele of a gene then number of different possible genotype = $\frac{n(n+1)}{2}$

Example of multiple allele :

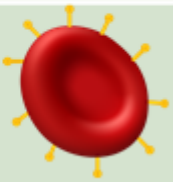
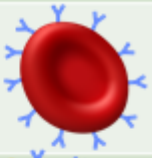
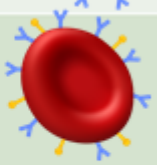
ABO blood group - ABO blood group system are determined by three alleles I^A, I^B, and i :–

I^A = dominant

I^B = dominant

i = recessive

Possible phenotypes - A, B, AB, O

Blood Group	Genotype	Antigen	Antibody	Picture
A	I^AI^A, I^AI^O	A	b	
B	I^BI^B, I^BI^O	B	a	
AB	I^AI^B	A & B	Neither a nor b	
O	I^OI^O	Neither A nor B	a & b	

Possible genotype number = $\frac{3(3+1)}{2} = 6$ genotype

Pleiotropic gene :– Gene which controls more than one character is called pleiotropic gene. This gene shows multiple phenotypic effect. We have so far seen the effect of a gene on a single phenotype or trait. There are however instances where a single gene can exhibit multiple phenotypic expression. Such a gene is called is the effect of a gene underlying mechanism of pleiotropy in most cases is the effect of a gene on metabolic pathways which contributes towards different phenotypes. An example of this is the disease phenylketonuria, which occurs in humans. The disease is caused by mutation in the gene that codes for the enzyme phenyl alanine hydroxylase

(single gene mutation). This manifests itself through phenotypic expression characterised by mental retardation and a reduction in hair and skin pigmentation.

Example :

1. **In Pea plant :-** Seed shape and size of starch grain control by same gene located on 7th chromosome. Occasionally, a single gene product may produce more than one effect. For example, starch synthesis in pea seeds is controlled by one gene. It has two alleles (B and b). Starch is synthesised effectively by BB homozygotes and therefore, large starch grains are produced. In contrast, bb homozygotes have lesser efficiency in starch synthesis and produce smaller starch grains. After maturation of the seeds, BB seeds are round and the bb seeds are wrinkled. Heterozygotes produce round seeds, and so B seems to be the dominant allele. But, the starch grains produced are of intermediate size in Bb seeds. So if starch grain size is considered as the phenotype, then from this angle, the alleles show incomplete dominance.

Therefore, dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype from this product as it does on the particular phenotype that we choose to examine, in case more than one phenotype is influenced by the same gene.

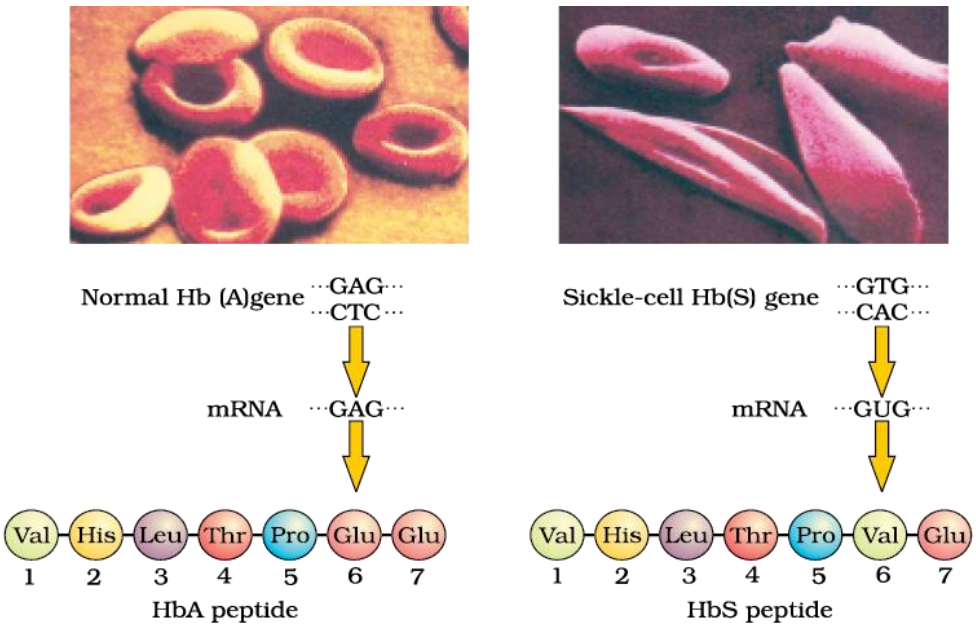
	Seed shape	Size of starch grain
BB	Round	Large
Bb	Round	Medium
Bb	Wrinkel	Small

Dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype from this product as it does on the particular phenotype that we choose to examine, in case more than one phenotype is influenced by the same gene.

2. **Sickle cell anaemia (Autosomal linked recessive) :-** Gene Hb^{S}_{β} provide a classical example of pleiotropy. It not only causes haemolytic anaemia but also results increased resistance to one type of malaria that caused by the parasite *Plasmodium falciparum*.

The sickle cell Hb^{S}_{β} allele also has pleiotropic effect on the development of many tissues and organs such as bone, lungs, kidney, spleen, heart. The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule. The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG.

The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure.



Micrograph of the red blood cells and the amino acid composition of the relevant portion of β-chain of haemoglobin: (a) From a normal individual; (b) From an individual with sickle-cell anaemia

This is an autosome linked recessive trait that can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous). The disease is controlled by a single pair of allele, Hb^A and Hb^S. Out of the three possible genotypes only homozygous individuals for Hb^S (Hb^SHb^S) show the diseased phenotype. Heterozygous (Hb^AHb^S) individuals appear apparently unaffected but they are carrier of the disease as there is 50 per cent probability of transmission of the mutant gene to the progeny, thus exhibiting sickle-cell trait. The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule. The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG. The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure.

Question-1: Father is heterozygous for A blood group and blood group of mother is AB. How many types of genotype and phenotypes are possible in this cross?

(1) 3 ,4

(2) 4, 3

(3) 5, 4

(4) 4, 5

Answer: (2) 4, 3

Question-2: What will be the phenotype and genotype of parental blood group of a family in which all four types of blood group are present in their children?

(1) Phenotype A, B
Genotype I^AI^O, I^BI^O

(2) Phenotype AB, B
Genotype I^AI^A, I^BI^O

(3) Phenotype A, A
Genotype I^AI^O, I^AI^A

(4) Phenotype A, B
Genotype I^AI^A, I^BI^B

Answer: (1) Phenotype A, B
Genotype I^AI^O, I^BI^O

Question-3: A man of 'A' blood group marries a woman of 'AB' blood group, which type of blood group in the child would indicate that the man is heterozygous for A ?

(1) A
(2) AB
(3) B
(4) O

Answer: (3) B

COMPLEMENTARY GENE, POLYGENIC INHERITANCE, CYTOPLASMIC INHERITANCE

COMPLEMENTARY GENE :

In this interaction two non allelic dominant gene regulate one character. For the development of dominant trait the dominant alleles of both genes must be present together. Absence of both dominant alleles of any genes will result in recessive expression.

e.g. :- Flower colour of lathyrous odoratus (sweet pea)

Flower colour is controlled by two genes 'C' and 'P'

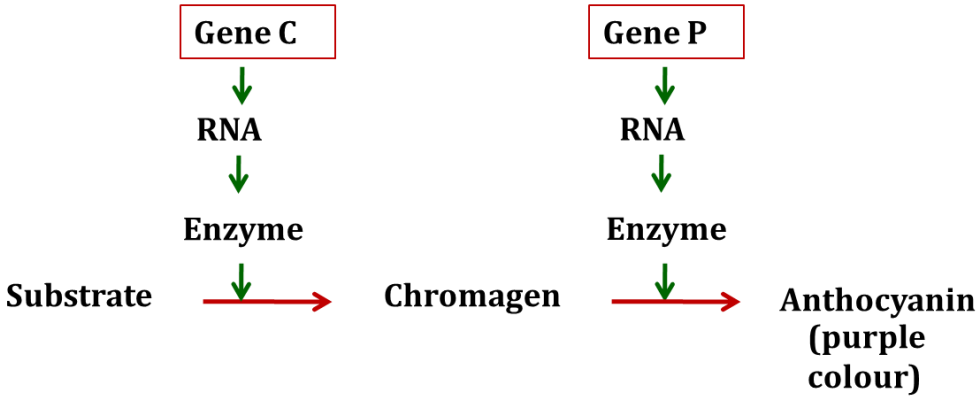
C_P_ :- Purple coloured.

C_ pp :- Colourless (white)

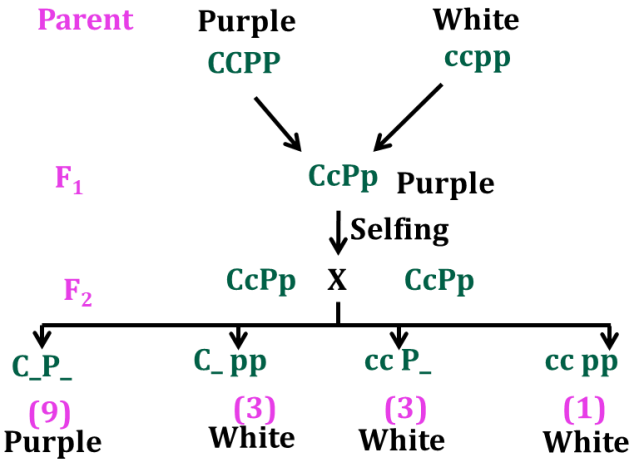
cc P_ :- Colourless (white)

cc pp :- Colourless (white)

Biochemical basis of colour production :-



	CP	Cp	cP	cp
CP	CCPP	CCPp	CcPP	CcPp
Cp	CCPp	CCpp	CcPp	Ccpp
cP	CcPP	CcPp	ccPP	ccPp
cp	CcPp	Ccpp	ccPp	ccpp



P.R. :- 9 : 7

G.R. :- 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

POLYGENIC INHERITANCE :

Mendel's studies mainly described those traits that have distinct alternate forms such as flower colour which are either purple or white. But if you look around you will find that there are many traits which are not so distinct in their occurrence and are spread across a gradient. For example, in humans we don't just have tall or short people as two distinct alternatives but a whole range of possible heights. Such traits are generally controlled by three or more genes and are thus called as polygenic traits.

Besides the involvement of multiple genes polygenic inheritance also takes into account the influence of environment. Human skin colour is another classic example for this.

In a polygenic trait the phenotype reflects the contribution of each allele, i.e., the effect of each allele is additive. Inheritance of characters in which one character is controlled by many genes and intensity of character depends upon the number of dominant alleles.

- **Number of phenotype = (2n + 1)**
- **Here n = number of polygene**

Example- kernal colour of wheat is regulated by two polygene

AABB (Red) X aa bb (White)

F_1 Aa Bb Intermediat

Phenotype F_2	1 :	4 :	6 :	4 :	1
	Full Red	Light Red	Intermediate Red	Very light Red	Full White
No of Dominant gene	4	3	2	1	0
	AABB	AaBB-2	AaBb-4	aaBb-2	aa bb
	1	AABb-2	aaBB-1 AAbb-1	Aabb-2	1

1	:	14	:	1
Full red		Intermediate		Full white

Example- :- Colour of the skin in Human.

- The inheritance of colour of skin in human was studied by **Devenport.**
- Human skin colour is regulated by three polygenes.
- To understand with the dominant forms A, B and C control skin colour in human with the dominant forms A, B, and C responsible for dark skin colour and the recessive forms a, b, and c for light skin colour.
- The genotype with all the dominant alleles (AABBCC) will have the darkest skin colour and that with all the recessive alleles (aabbcc) will have the lightest skin colour. As expected the genotype with three dominant alleles and three recessive alleles will have an intermediate skin colour. In this manner the number of each type of alleles in the genotype would determine the darkness or lightness of the skin in an individual.
- When a Negro Black (AA BB CC) phenotype is crossed with white (aa bb cc) phenotype, intermediate phenotype is produced in F1 generation.

Negro Black
AA BB CC
X
White
aa bb cc

↓

Aa Bb Cc → F_1 - generation
[Intermediate brown/mullato]

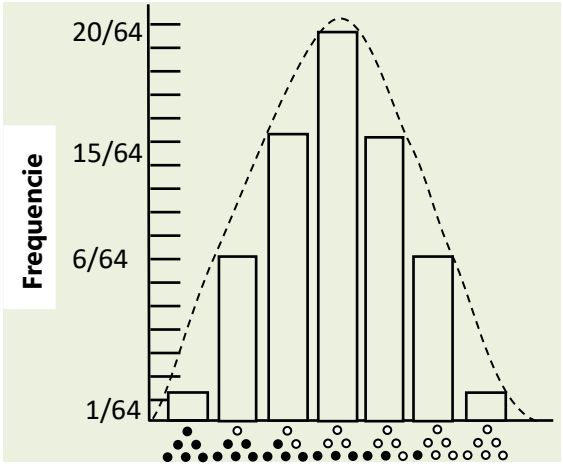
↓ (Inbreeding)

F_2-generation

↓

Phenotypic ratio will be :-

Negro Black	:	Very Drak Brown	:	Dark Brown	:	Mullato	:	Light Brown	:	Very Light Brown	:	White
1	:	6	:	15	:	20	:	15		6		1



Mendel's studies mainly described those traits that have distinct alternate forms such as flower colour which are either purple or white. But if you look around you will find that there are many traits which are not so distinct in their occurrence and are spread across a gradient. For example, in humans we don't just have tall or short people as two distinct alternatives but a whole range of possible heights. Such traits are generally controlled by three or more genes and are thus called as polygenic traits. Besides the involvement of multiple genes polygenic inheritance also takes into account the influence of environment. Human skin colour is another classic example for this. In a polygenic trait the phenotype reflects the contribution of each allele, i.e., the effect of each allele is additive.

CYTOPLASMIC INHERITANCE

- **Discovered by Correns.**
- Inheritance of characters which are controlled by **cytogene** or cytoplasm is called cytoplasmic inheritance.
- Genes which are present in cytoplasm called 'cytogene' or 'plasmagene' or extra nuclear gene. Total cytogene present in cytoplasm is called 'Plasmon'
- A gene which is located in the nucleus is called ' **karyogene'.**
- Inheritance of cytogene in organisms occurs only through the female. Because female gamete has karyoplasm, simultaneously it has cytogene because of more cytoplasm.
- The male gamete of higher plant is called male nucleus. It has very minute [equivalent to nil] cytoplasm. so male gamete only inherited karyogene.
- Thus, inheritance of cytogene occurs only through female. **(also called maternal inheritance)**
- If there is a reciprocal cross in this condition, then results may be affected.
- Cytoplasmic inheritance involving essential organelles like, Chloroplast and mitochondria called as **organellar inheritance.**

Question-1: A quantitative character is controlled by four polygene's. Find out probability of parental plant in F_2 generation?

(1) 4/256
(2) 2/256
(3) 8/256
(4) 1/256

Answer: (2) 2/256

Question-2: **In F_2 generation of A quantitative character 17 phenotypic categories are obtained. This character is controlled by how many polygene's?**

(1) 4
(2) 2
(3) 8
(4) 16

Answer: **(3) 8**

Question-3: **A plant is homozygous for A and B alleles have 200 cm height while the minimum height of plant is 120 cm. find out height of Aabb genotypic plant. :-**

(1) 120
(2) 140
(3) 160
(4) 180

Answer: **(2) 140**

Question-4: **A plant with genotype TtRrGg produce fruit of weight 300 gm while the minimum weight of fruit is 210 gm. find out Max. weight of fruit. :-**

(1) 390
(2) 330
(3) 360
(4) 380

Answer: **(1) 390**

Question-5: **With respect to flower colour of *Lathyrous odoratous* genotype of a plant is CcPp. This plant is crossed with Ccpp genotypic plant. Find out probability of purple and white flower respectively in offspring's?**

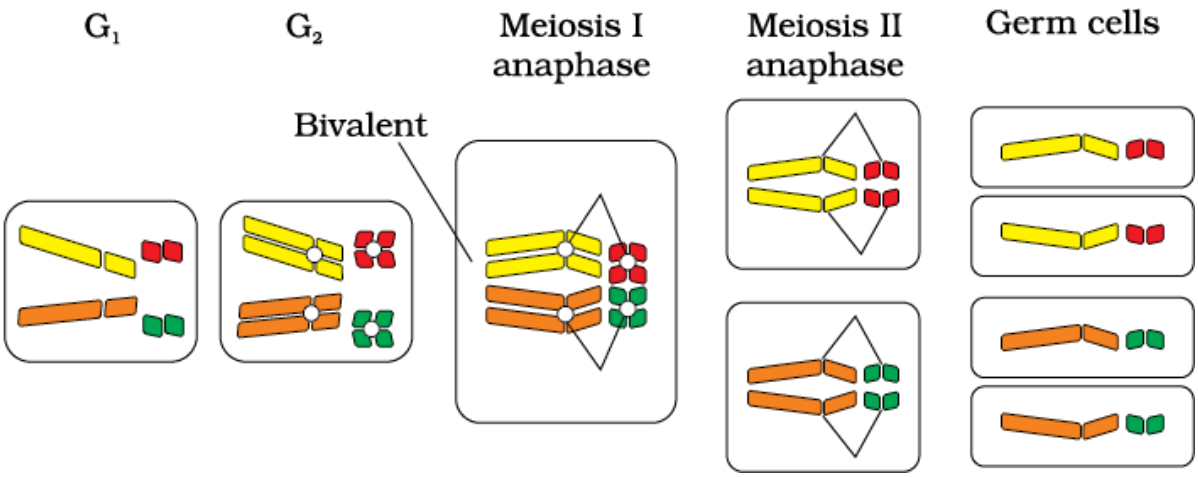
(1) 3, 5
(2) 5, 3
(3) 2, 6
(4) 6, 2

Answer: **(1) 3, 5**

CHROMOSOMAL THEORY OF INHERITANCE :

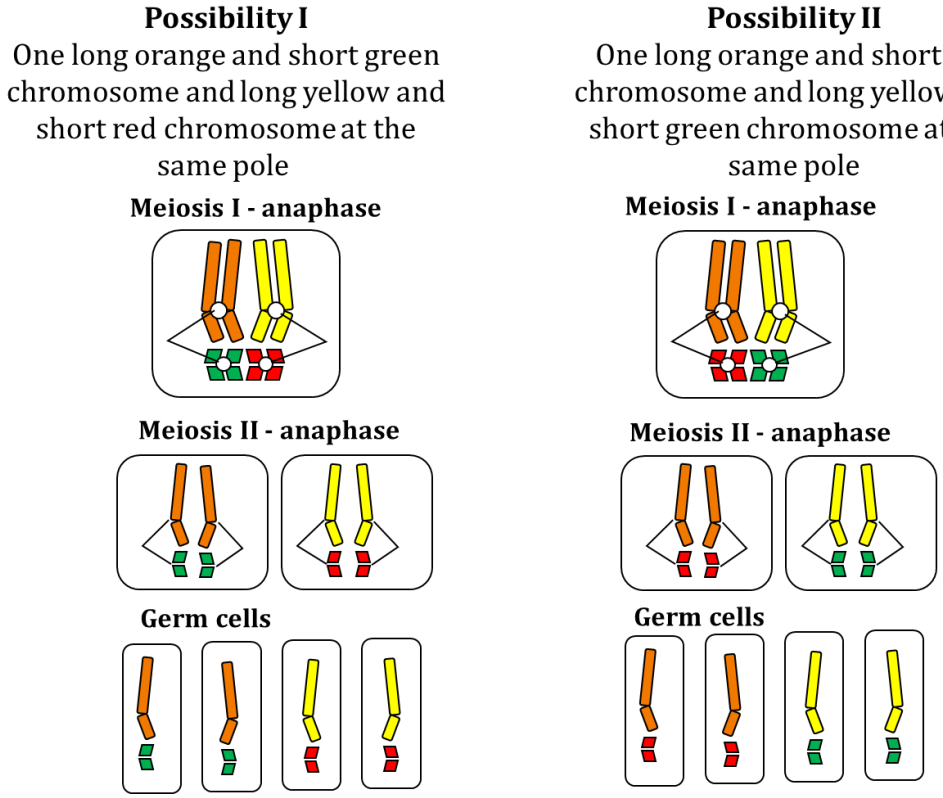
- This theory was proposed by **Walter Sutton and Theodor Boveri (1902).**
- In 1900, three Scientists (de Vries, Correns and von Tschermak) independently rediscovered Mendel's results on the inheritance of characters. Also, by this time due to advancements in microscopy that were taking place, scientists were able to carefully observe cell division. This led to the discovery of structures in the nucleus that appeared to double and divide just before each cell division. These were called chromosomes (coloured bodies, as they were visualised by staining). By 1902, the chromosome movement during meiosis had been worked out.
- **Walter Sutton and Theodore Boveri noted that the behaviour of chromosomes was parallel to the behaviour of genes and used chromosome movement to explain Mendel's laws.**

- Recall that you have studied the behaviour of chromosomes during mitosis (equational division) and during meiosis (reduction division).The important things to remember are that chromosomes as well as genes occur in pairs. The two alleles of a gene pair are located on homologous sites on homologous chromosomes.



Meiosis and germ cell formation in a cell with four chromosomes. You can see how chromosomes segregate when germ cells are formed

- During metaphase of meiosis I, the two chromosome pairs can align at the metaphase plate independently of each other.
- To understand this, compare the chromosomes of four different colour in the left and right columns. In the left column (Possibility I) orange and green is segregating together. But in the right-hand column (Possibility II) the orange chromosome is segregating with the red chromosomes


Possibility I
One long orange and short green chromosome and long yellow and short red chromosome at the same pole
Meiosis I - anaphase
Meiosis II - anaphase
Germ cells
Possibility II
One long orange and short red chromosome and long yellow and short green chromosome at the same pole
Meiosis I - anaphase
Meiosis II - anaphase
Germ cells

Independent Assortment of Chromosomes

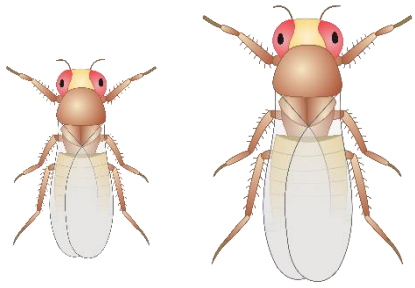
A Comparison between the Behaviour of Chromosomes and Genes

A (Gene)	B (Chromosome)
Occur in pairs	Occur in pairs
Segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete	Segregate at gamete formation and only one of each pair is transmitted to a gamete
Independent pairs segregate independently of each other	One pair segregates independently of another pair

MORGAN AND DROSOPHILA

Sutton and Boveri argued that the pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factors they carried. Sutton united the knowledge of chromosomal segregation with Mendelian principles and called it the chromosomal theory of inheritance.

Following this synthesis of ideas, experimental verification of the chromosomal theory of inheritance by Thomas Hunt Morgan and his colleagues, led to discovering the basis for the variation that sexual reproduction produced. Morgan worked with the tiny fruit files, *Drosophila melanogaster*, which were found very suitable for such studies as: -



Drosophila melanogaster
(a) Male (b) Female

1. They could be grown on simple synthetic medium in the laboratory.
2. They complete their life cycle in about two weeks.
3. A single mating could produce a large number of progeny flies.
4. There was a clear differentiation of the sexes the male and female flies are easily distinguishable.
5. It has many types of hereditary variations that can be seen with low power microscopes.

In 1900, three Scientists (de Vries, Correns and von Tschermak) independently rediscovered Mendel's results on the inheritance of characters. Also, by this time due to advancements in microscopy that were taking place, scientists were able to carefully observe cell division. This led to the discovery of structures in the nucleus that appeared to double and divide just before each cell division. These were called chromosomes (colored bodies, as they were visualised by staining). By 1902, the chromosome movement during meiosis had been worked out. Walter Sutton and Theodore Boveri noted that the behaviour of chromosomes was parallel to the behaviour of genes and used chromosome movement to explain Mendel's laws. Recall that you have studied the behaviour of chromosomes during mitosis (equational division) and during meiosis (reduction division). The important things to remember are that chromosomes as well as genes occur in pairs. The two alleles of a gene pair are located on homologous sites on homologous chromosomes

.

Question-1: Who proposed chromosomal theory of inheritance: -

(1) Mendel
(2) Morgan
(3) Sutton and Boveri
(4) Correns

Answer: (3) Sutton and Boveri

Question-2: Chromosomal theory of inheritance proposed in: -

(1)1900
(2) 1902
(3) 1905
(4)1865

Answer: (2) 1902

Question-3: Which statement is true for gene: -

(1) Occur in pairs
(2) Segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete
(3) Independent pairs segregate independently of each other
(4) All of above

Answer: (4) All of above

Question-4: Morgan found drosophila suitable for genetics studies because :-

(1) They could be grown on simple synthetic medium in the laboratory.
(2) They complete their life cycle in about two weeks.
(3) A single mating could produce a large number of progeny flies
(4) All of above

Answer: (4) All of above

Question-5: Chromosomal theory of inheritance experimentally proofed by :-

(1) T.H. Morgan
(2) Gregor Mendel
(3) Sutton and Boveri
(4) Carl Correns

Answer: (1) T.H. Morgan

Question-6: Genome of *Drosophila melanogaster* is :-

(1) 2n = 8
(2) 2n = 4
(3) n = 8
(4) n = 4

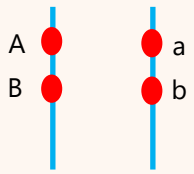
Answer: (1) 2n = 8

LINKAGE AND RECOMBINATION :

LINKAGE :- The physical association of genes on a chromosome is called linkage. Collective inheritance of character is due to linkage. Sex linkage was first discovered by Morgan in Drosophila and coined the term linkage. He proposed the theory of linkage.

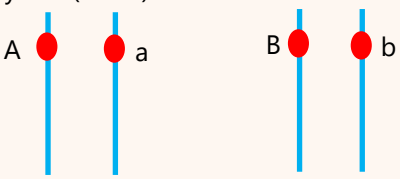
- Linkage and independent assortment can be represented in dihybrid plant, as

In case of linkage in dihybrid (AaBb)



In produces two types of gamete
AB : ab

In case of independent assortment in dihybrid (AaBb)



In produces four types of gamete
AB : ab : aB : Ab

THEORY OF LINKAGE :

- Linked genes are linearly located on same chromosome. They get separated if exchange (crossing over), takes place between them.
- **Linkage group:-** All the genes which are located on one pair of homologous chromosome form one linkage group.
- Genes which are located on homologous chromosomes inherit together so we consider one linkage group.
- **Number of Linkage group = Number of homologous chromosome pairs or Haploid no. of chromosomes.**

	2n	n	Pair	Linkage group
Pea	14	7	7	7
Maize	20	10	10	10
Drosophila	8	4	4	4
Barley	14	7	7	7
Mouse	42	21	21	21

- **Strength of linkage α 1/ distance between the genes.** It means, if the distance between two genes is increased then strength of linkage is reduced and it proves that greater is the distance between genes, the greater is the probability of their crossing over.
- Crossing over obviously disturbs or degenerates linkage. Linked genes can be separated by crossing over.

FACTORS AFFECTING CROSSING OVER (C.O.) :

(1) Distance ↑ = C.O. ↑

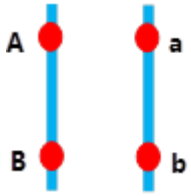
(2) Age ↑ = C.O. ↓

(3) Sex - Male C.O. ↓ (Crossing over is totally absent in male Drosophila)

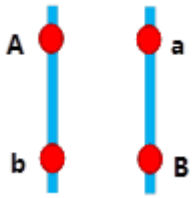
ARRANGEMENT OF LINKED GENES ON CHROMOSOMES :

The arrangement of linked genes in any dihybrid is of two types.

[a] Cis – Arrangement :- When, two dominant genes are located on one chromosome and both recessive genes located on another chromosome, such type of arrangement is termed as cis-arrangement. Cis-arrangement is an original arrangement. Two types of gametes can be produced in cis-arrangement -(AB) and (ab).



[b] Trans-arrangement :- When a chromosome bears one dominant and one recessive gene, and another chromosome also possess one dominant and one recessive gene, such type of arrangement is called trans-arrangement. Trans-arrangement is not an original form. It is due to crossing over. Two types of gamete also formed in trans-arrangement but it is different from cis-arrangement- (Ab) and (aB).



TYPES OF LINKAGE: - There are two types of linkage

1. COMPLETE LINKAGE :

- Linkage in which genes always show parental combination. It never forms new combination.
- Crossing over is absent in it. Such genes are located very close on the chromosomes.
- Such type of linkage is very rare in nature. e.g., male Drosophila, female silk moth.

2. INCOMPLETE LINKAGE :

- When new combinations (<50%) also appear along with parental combination in off spring, this type of linkage is called incomplete linkage.
- The new combinations are formed due to crossing over.

  Morgan's student Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and mapped their position on the chromosome.

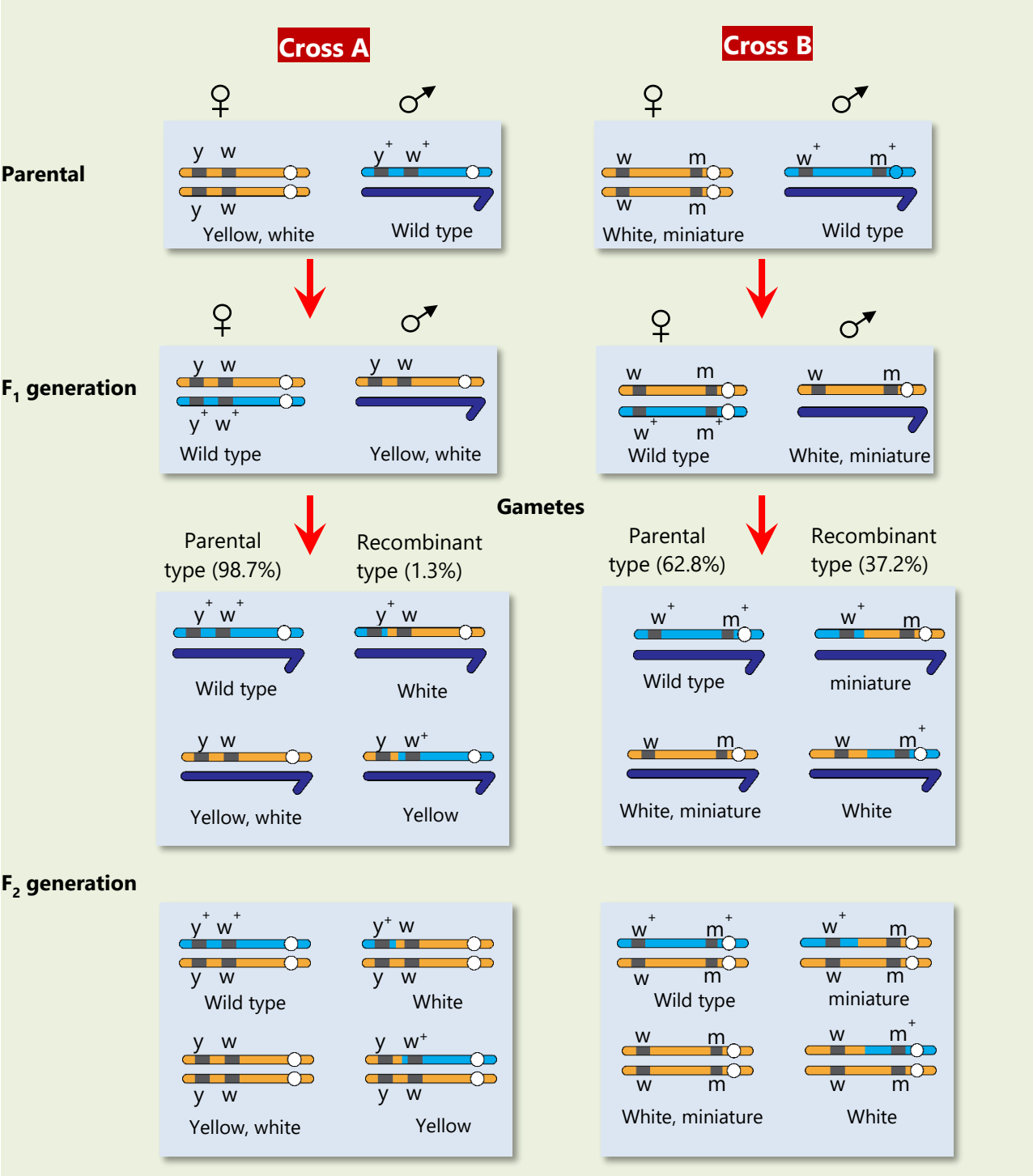


Figure : Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between geney and w; Cross B shows crossing between genes w and m. Here dominant wild type alleles are represented with (+) sign in superscript.

Note :- The strength of linkage between y and w is higher than w and m.

Morgan carried out several dihybrid crosses in Drosophila to study genes that were sex-linked. The crosses were similar to the dihybrid crosses carried out by Mendel in peas. For example Morgan hybridised yellow-bodied, white-eyed females to brown-bodied, red-eyed males and intercrossed their F_1 progeny. He observed that the two genes did not segregate independently of each other and the F_2 ratio deviated very significantly from the 9:3:3:1 ratio (expected when the two genes are independent).

Question-1: An exception to the law of independent assortment is :-

(1) Dominance
(2) Incomplete dominance
(3) Segregation
(4) Linkage

Answer: (4) Linkage

Question-2: Maize has 10 pairs of chromosomes. How many linkage groups are present :-

(1) 05
(2) 10
(3) 20
(4) 40

Answer: (2) 10

Question-3: Genotype of plant is $\frac{++}{ab}$ in which genes show 20% recombination. Find out percentage of gametes which contains '+b' genotype?

(1) 10%
(2) 20%
(3) 30%
(4) 40%

Answer: (1) 10%

Question-4: Genotype of plant is $\frac{+r}{d+}$ in which genes show 30% recombination. Find out percentage of gametes which contains '++' genotype :-

(1) 10%
(2) 15%
(3) 20%
(4) 25%

Answer: (2) 15%

Question-5: Experimental proof for sex-linked gene was given by :-

(1) Morgan
(2) Muller
(3) Mendel
(4) Johannsen

Answer: (1) Morgan

Question-6: If Mendel has chosen to study traits determined by linked genes he would not have discovered :-

(1) Law of segregation
(2) Law of dominance
(3) Law of independent assortment
(4) Law of unit character

Answer: (3) Law of independent assortment

SEX LINKED INHERITANCE :

When the genes are present on sex-chromosome is termed as sex linked gene and such phenomenon is known as sex-linkage.

TYPES OF SEX LINKAGE :

1. X-LINKAGE :-

Genes of many somatic characters are found on X-chromosome. The inheritance of x-linked character may be through the males and females.

Example of X-linkage :-

[i] Eye colour in Drosophila :- Eye colour in Drosophila is controlled by a X-linked gene. If a red eye colour gene is represented as '+' and white eyed colour represented as 'w', then on basis of this different type of genotypes are found in Drosophila.

Gene for red eye is dominant (+) and white colour of eye is recessive (w)

Homozygous red eyed female = $X^+ X^+$

Heterozygous red eyed female = $X^+ X^w$

Homozygous white eyed female = $X^w X^w$

Hemizygous red eyed male = $X^+ Y$

Hemizygous white eyed male = $X^w Y$

It is clear by above different types of genotype that female is either homozygous or heterozygous for eye colour. But, for the male eye colour, it is always hemizygous.

[ii] Haemophilia :-Haemophilia is also called "bleeder's disease" and first discovered by **John Otto (1803).** The gene of haemophilia is recessive.

On the basis of x-linked, following types of genotype are found.

$X^h X$ = Carrier female

$X^h X^h$ = Affected female

$X^h Y$ = Affected male.

Haemophilia –A → due to lack of factor -VIII (Antihaemophilic globulin AHG). It shows lethality in homogygous condition.

Haemophilia B or Christmas disease → due to lack of factor - IX (Plasma thromboplastin component)

[iii] Colour Blindness :- The inheritance of colour-blindness is alike as haemophilia, but it is not a lethal disease so it is found in male and female.(discovered by Horner) Types of X-linked colour blindness are :-

[a] Protanopia :- It is for red colour.

[b] Deuteranopia :- It is for green colour

❖ Colour blindness is checked by ishihara - chart.

It is a sex-linked recessive disorder due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour. This defect is due to mutation in certain genes present in the X chromosome. it occurs in about 8 percent of males and only about 0.4 percent of females. This is because the genes that lead to red-green colour blindness are on the X chromosome. Males have only one X chromosome and females have two. The son of a woman who carried the gene has a 50 percent chance of being colour blind. The mother is not herself colour blind because the gene is recessive. That means that its effect is suppressed by her matching dominant normal gene. A daughter will not normally be colour blind, unless her mother is a carrier and her father is colour blind.

Summary :-
Haemophilia : This sex linked recessive disease, which shows its transmission from unaffected carrier female to some of the male progeny has been widely studied. In this disease, a single protein that is a part of the cascade of proteins involved in the clotting of blood is affected. Due to this, in an affected individual a simple cut will result in non-stop bleeding. The heterozygous female (carrier) for haemophilia may transmit the disease
to sons. The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic (unviable in the later stage of life). The family pedigree of Queen Victoria shows a number of haemophilic descendants as she was a carrier of the disease.

Other examples of X-linkage

[iv] Diabetes insipidus (recessive).

[v] Duchenne muscular dystrophy (recessive).

[vi] Pseudoricketes (Dominant)

[vii] Defective enamel of teeth (Dominant)

2. Y- LINKAGE -

The genes of some somatic characters are located on Y- chromosome. The inheritance of such type of character is only through the males. Such type of character is called Holandric character. These characters are found only in male.

e.g. (1) Gene which forms TDF /sry-gene

(2) Hypertrichosis (excessive hair on ear pinna.)

Gene which is located on differential region of Y - chromosome is known as Holandric gene.

APPLICATION OF LINKAGE :

Distance between linked genes can be identified by the incomplete linkage. Its unit is centi Morgan.

$$\text{Strength of linkage} \propto \frac{1}{\text{Distance b/w linked gene}} \propto \frac{1}{\text{Crossing Over}}$$

Genetic map/Linkage map/chromosome map - In genetic map different genes are linearly arranged according to % of recombination (μ Distance) between them. With the help of genetic map, we can find out the position of a particular gene on chromosome. Genetic map is helpful in the study of genome.

TYPES OF INHERITANCE OF SEX-LINKED CHARACTERS: -

1. CRISS CROSS INHERITANCE (MORGAN) :-

In criss-cross inheritance male or female parent transfer a X- linked character to grandson or granddaughter through the offspring of opposite sex.

(a) **Diagenic (Diagynic) :-** Inheritance in which characters are inherited from father to the daughter and from daughter to grandson.

Father →daughter→ grandson.

(b) **Diandric :-** Inheritance in which characters are inherited from mother to the son and from son to granddaughter.
Mother →Son → Grand-daughter.

2. Non criss-cross inheritance :-

In this inheritance male or female parent transfer sex linked character to grandson or granddaughter through the offspring of same sex.

(a) Hologenic (Hologynic) :- Mother→ Daughter →Grand-daughter (female to female)

(b) Holandric :- Father →Son →Grand-son (male to male)

SEX-LIMITED CHARACTER

These characters are present in one sex and absent in another sex. But their genes are present in both thesexes and their expression is depend on sex hormone.

Example: - **Secondary sexual characters** → these genes are located on the autosomes and these genes are present in both male and female, but effect of these are dependent upon presence or absence of sex-hormones. For example - genes of beard-moustache express their effects only in the presence of male hormone testosterone.

SEX INFLUENCED CHARACTERS

Genes of these characters are also present on autosomes but they are influenced differently in male and female. In heterozygous condition their effect is different in both the sexes.

Example: -**Pattern baldness :-** Gene of pattern baldness is dominant (B)

Genotype	Male	Female
BB	Baldness present	Baldness present
Bb	Baldness present	Baldness absent
bb	Baldness absent	Baldness absent

Gene Bb shows partiality in male and female, Baldness is found in male due to effect of this gene, but baldness is absent in female with this genotype.

Question-1: A heterozygous red eyed female Drosophila, is crossed with hemizygous red eyed male Drosophila. Find out percentage of red eyed female offspring's?

(1) 100

(2) 50

(3) 75

(4) 25

Answer: (1) 100

Question-2: X-linked recessive gene easily express in :-

(1) Male

(2) Female

(3) Equal in male and female

(4) Not easily express

Answer: (1) Male

Question-3: This disease falls under the same category as colorblindness in man :-

(1) Presbyopia

(2) Night blindness

(3) Diabetes insipidus

(4) Haemophilia

Answer: (4) Haemophiia

Question-4: There are three genes a, b, c percentage of crossing over between a and b is 20%, b and c is 28% and a and c is 8%. What is the sequence of genes on chromosome?

(1) b, a, c

(2) a, b, c

(3) a, c, b

(4) None

Answer: (1) b, a, c

Question-5: The recombination frequency between linked genes are :-

DC = 25 % **DB =35 %** **DA = 40 %** **CB = 10 %**

CA = 15 % **AB = 5 %**

(1) ABCD

(2) BACD

(3) CADB

(4) DBAC

Answer: (1) ABCD

Question-6: Result of test cross in Drosophila is :-

++/ab = 41 **ab/ab = 41** **+b/ab = 9** **a+/ab = 9**

Find out :- % of recombination

(1) 82%

(2) 18%

(3) 36%

(4) 64%

Answer: (2) 18%

Question-7: Result of test cross in a plant is :-

++/ab = 202 ab/ab = 198 +b/ab = 798 a+/ab = 802

Find out :- % of recombination

(1) 80%

(2) 30%

(3) 70%

(4) 20%

Answer: (4) 20%

SEX DETERMINATION :

Establishment of sex through differential development in an individual at an early stage of life, is called sex determination. There are different methods for sex determination in organisms like allosomic sex determination, haplodiploidy, etc.

SEX DETERMINATION ON THE BASIS OF FERTILIZATION :

Three types :

1. **PROGAMIC :-** Sex is determined before fertilization.
   e.g. - drone in honey bee
2. **SYNGAMIC :-** Sex is determined during fertilization.
   e.g. - most of plants & animals
3. **EPIGAMIC :-** Sex is determined after fertilization.
   e.g. - Female in honey bee

MECHANISM OF SEX DETERMINATION :

[1] ALLOSOMIC DETERMINATION OF SEX :

Chromosomes are of two types –

(a) Autosomes or somatic chromosomes - These regulate somatic characters.

(b) Allosomes or Heterosomes or Sex chromosomes –

These chromosomes are associated with sex determination. Term "Allosome" & "Heterosome" were given by **Montgomery**.

Sex chromosomes were discovered by **"Mc Clung"** in grass hopper

X- Chromosome was discovered by **"Henking"** and called it 'x-body'.

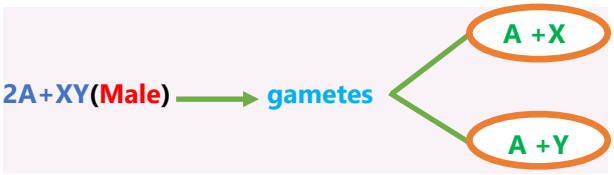
Wilson & Stevens proposed chromosomal theory for sex determination.

(1) XX - XY TYPE OR LYGAEUS TYPE : -

This type of sex determination first observed by **Wilson & Stevens** in Lygaeus insect. Two types-

(a) XX female and XY male :- In this type of sex determination female is Homogametic i.e produces only one type of gamete.

- Male is heterogametic (male produces two types of gamete)



e.g. Man

(b) ZW female and ZZ male :- In this type of sex determination female is Heterogametic i.e produces two types of gamete and male individual is homogametic i.e produces one type of gamete. It is found in **birds.**

(2) **XX female and XO male :- or "Protenor type" :-** In this type of sex determination there is deficiency of one chromosome in male. In this type, female is homogametic and male is heterogametic.

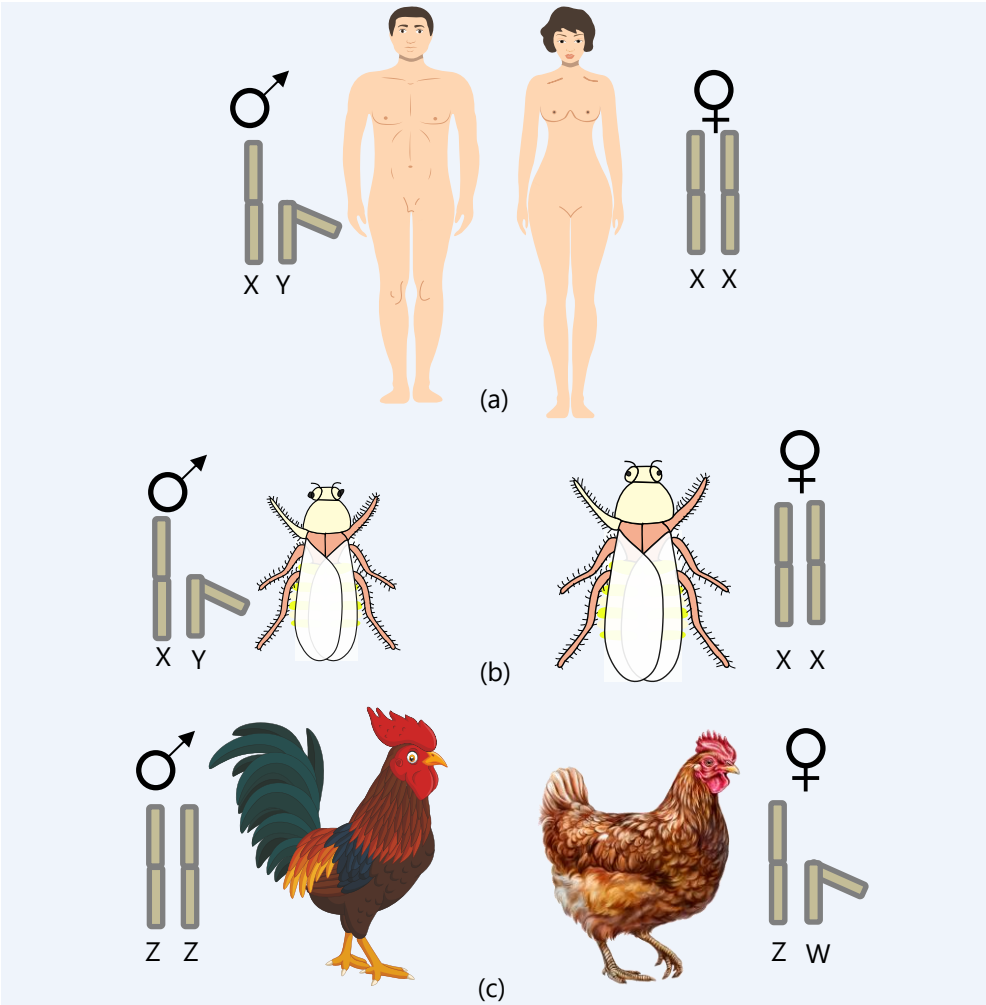
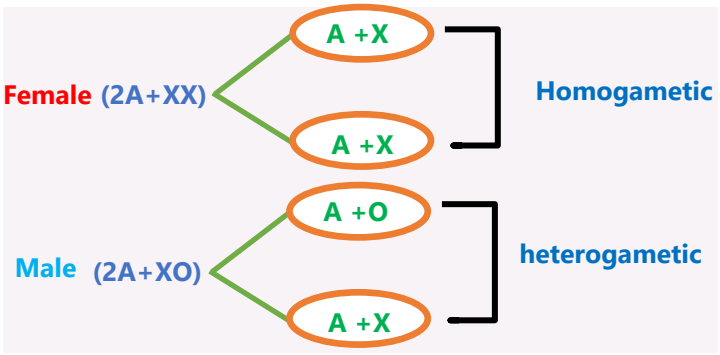


Figure : Determination of sex by chromosomal differences: (a, b) Both in humans and in Drosophila, the female has a pair of XX chromosomes (homogametic) and the male XY (heterogametic) composition; (c) In many birds, female has a pair of dissimilar chromosomes ZW and male two similar ZZ chromosomes.



Example :

1. Grass hopper
2. Cockroach

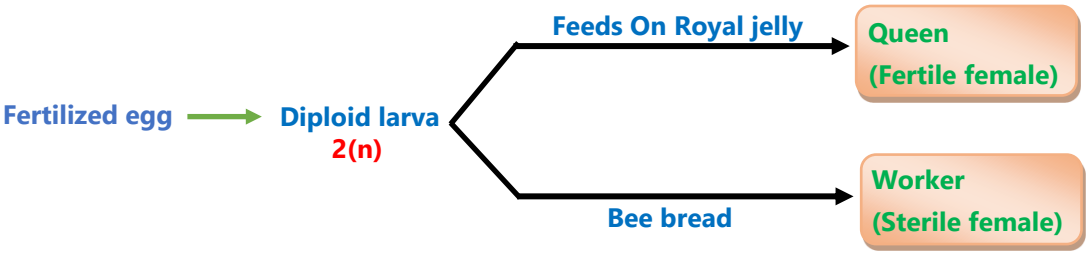
HAPLOID - DIPLOID MECHANISM (SEX DETERMINATION IN HONEY BEE)

Sex determination takes place by sets of chromosomes.

Diploid (two sets) → **Female**

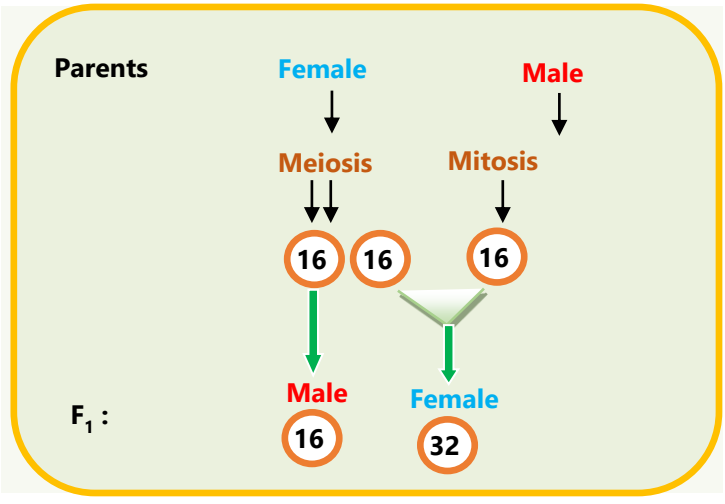
Haploid (One set) → **Male**

In honey bee, male individual (Drone) develops from unfertilized eggs (Haploid). Male develop by parthenogenesis. Queen and worker bees develop from diploid eggs *i.e.* fertilized egg.



The sex determination in honey bee is based on the number of sets of chromosomes an individual receives. An offspring formed from the union of a sperm and an egg develops as a female (queen or worker), and an unfertilized egg develops as a male (drone) by means of parthenogenesis. This means that the males have half the number of chromosomes than that of a female.

The females are diploid having 32 chromosomes as haplodipoloid sex-determination system and has special characteristic features such as the males produce sperms by mitosis (figure), they do not have father and thus cannot have sons, but have a grandfather and can have grandsons.



Summary :-

It has already been mentioned that the sex determining mechanism in case of humans is XY type. Out of 23 pairs of chromosomes present, 22 pairs are exactly same in both males and females; these are the autosomes. A pair of X-chromosomes are present in the female, whereas the presence of an X and Y chromosome are determinant of the male characteristic. During spermatogenesis among males, two types of gametes are produced. 50 per cent of the total sperm produced carry the X-chromosome and the rest 50 per cent has Y-chromosome besides

the autosomes. Females, however, produce only one type of ovum with an X-chromosome. There is an equal probability of fertilisation of the ovum with the sperm carrying either X or Y chromosome. In case the ovum fertilises with a sperm carrying X-chromosome the zygote develops into a female (XX) and the fertilisation of ovum with Y-chromosome carrying sperm results into a male offspring. Thus, it is evident that it is the genetic makeup of the sperm that determines the sex of the child. It is also evident that in each pregnancy there is always 50 per cent probability of either a male or a female child. It is unfortunate that in our society women are blamed for giving birth to female children and have been ostracised and ill-treated because of this false notion.

Question-1: The machanism of sex determination in birds shows :-

(1) Male heterogametic
(2) Both heterogametic
(3) Female heterogametic
(4) Both homogametic

Answer: (3) Female heterogametic

Question-2: How the sex of offsprings determined in humans :-

(1) Sex chromosome of mother
(2) Size of ovum
(3) Size of sperm
(4) Sex chromosome of father

Answer: (4) Sex chromosome of father

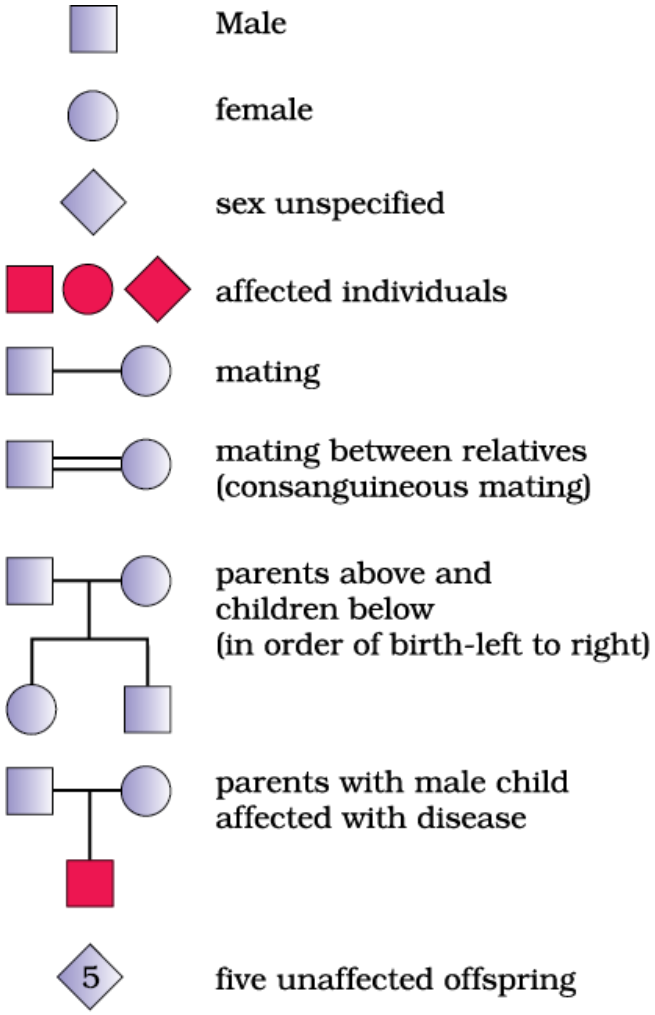
Question-3: Which chromosome set is found in male grasshopper :-

(1) 2A + XY
(2) 2A + XO
(3) 2A + YY
(4) 2A + XX

Answer: (2) 2A + XO

PEDIGREE ANALYSIS :

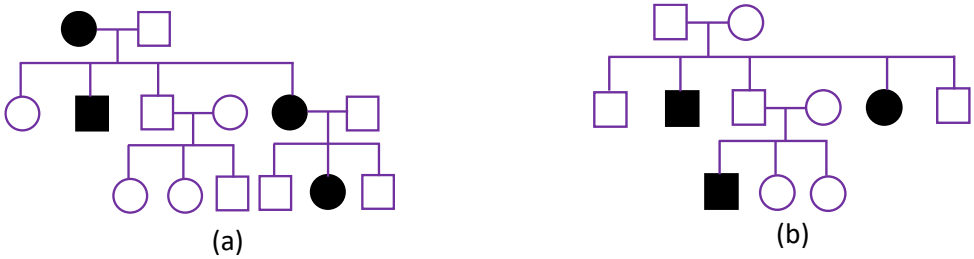
Study of ancestoral history of man of transmission of genetic characters from one generation to next, is pedigree analysis. Dwarfism, albinism, colour blindness, haemophilia etc. are genetically transmitted characters. To study and analyse them a pedigree of genetic facts/data and following symbols are used:



Pedigree analysis provides valuable informations regarding genetical make up of human beings. If any genetic disease is occurring in a family, then pedigree analysis provides guidance to forthcoming parents about their future progenies for example- polydactyly in humans.

A representative pedigree is shown in Figure for dominant and recessive traits, discuss with your teacher and design pedigrees for characters linked to both autosomes and sex chromosome.

Examples :-



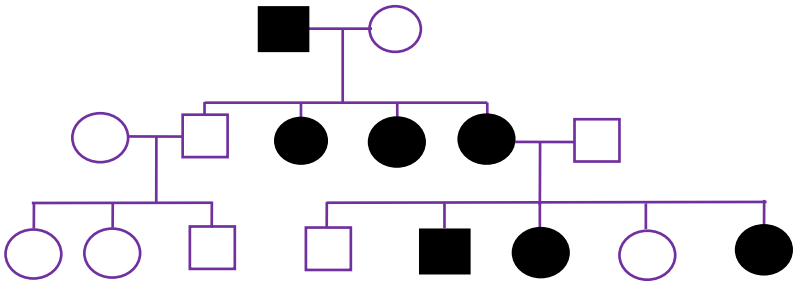
Representative pedigree analysis of (a) Autosomal dominant trait (Mytotonic Dystrophy (b) Autosomal recessive trait (Sickle Cell Anaemia)

Summary :-
The idea that disorders are inherited has been prevailing in the human society since long. This was based on the heritability of certain characteristic features in families. After the rediscovery of Mendel's work the practice of analysing inheritance pattern of traits in human beings began. Since it is evident that control crosses that can be performed in pea plant or some other organisms, are not possible in case of human beings, study of the family history about inheritance of a particular trait provides an alternative. Such an analysis of traits in a several of generations of a family is called the pedigree analysis.

Question-1: Study the given pedigree carefully

(1) AR
(2) AD
(3) XR
(4) XD

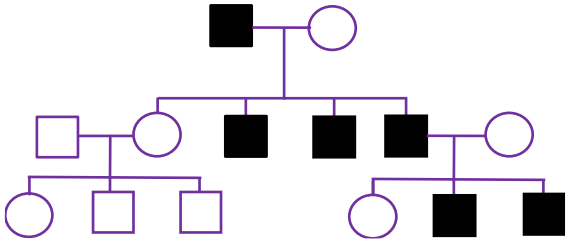
Answer: (4) XD



Question-2: Given pedigree show the inheritance of :-

(1) Y-linked traits
(2) X- linked traits
(3) Autosomal dominant
(4) Autosomal recessive

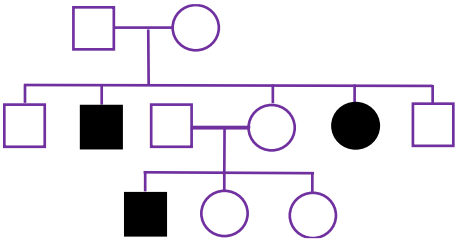
Answer: (1) Y-linked traits



Question-3: Given pedigree show the inheritance of :- (NCERT)

(1) AR
(2) AD
(3) XR
(4) XD

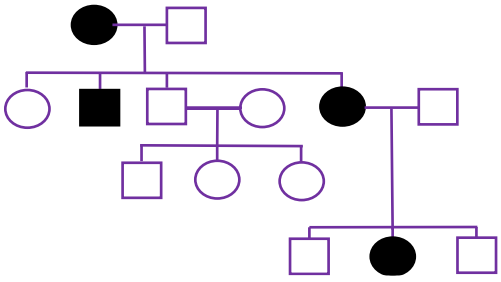
Answer: (1) AR



Question-4: Given pedigree show the inheritance of :- (NCERT)

(1) AR
(2) AD
(3) XR
(4) XD

Answer: (2) AD



MENDELIAN DISORDERS

Broadly, genetic disorders may be grouped into two categories – Mendelian disorders and Chromosomal disorders. Mendelian disorders are mainly determined by alteration or mutation in the single gene. These disorders are transmitted to the offspring on the same lines as we have studied in the principle of inheritance. The pattern of inheritance of such Mendelian disorders can be traced in a family by the pedigree analysis. Most common and prevalent Mendelian disorders are Haemophilia, Cystic fibrosis, Sickle cell anaemia, Colour blindness, Phenylketonuria, Thalassemia, etc. It is important to mention here that such Mendelian disorders may be dominant or recessive. By pedigree analysis one can easily understand whether the trait in question is dominant or recessive. Similarly, the trait may also be linked to the sex chromosome as in case of haemophilia. It is evident that this X-linked recessive trait shows transmission from carrier female to male progeny.

SEX LINKED MENDELIAN DISORDER

(1) Colour Blindness : It is a **sex-linked recessive disorder** due to defect in either red or green cone of eye resulting in failure to discriminate between red and green colour. This defect is due to mutation in certain genes present in the X chromosome. It occurs in about 8 per cent of males and only about 0.4 per cent of females. This is because the genes that lead to red-green colour blindness are on the X chromosome. Males have only one X chromosome and females have two. The son of a woman who carries the gene has a 50 per cent chance of being colour blind. The mother is not herself colour blind because the gene is recessive. That means that its effect is suppressed by her matching dominant normal gene. A daughter will not normally be colour blind, unless her mother is a carrier and her father is colour blind.

(2) Haemophilia : This **sex-linked recessive disease**, which shows its transmission from unaffected carrier female to some of the male progeny has been widely studied. In this disease, a single protein that is a part of the cascade of proteins involved in the clotting of blood is affected. Due to this, in an affected individual a simple cut will result in non-stop bleeding.

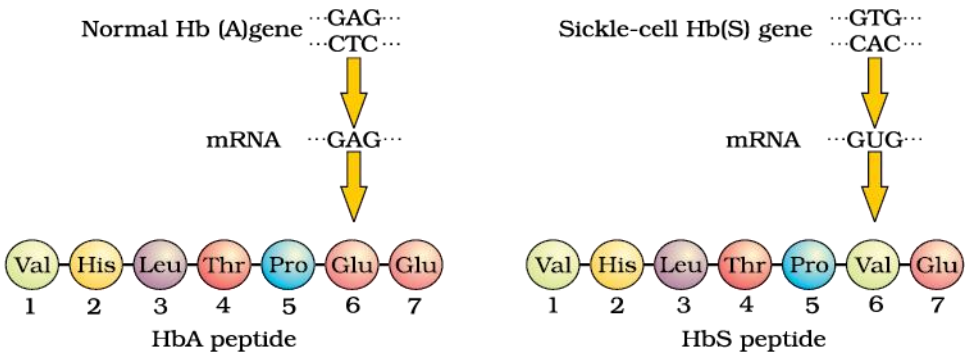
The heterozygous female (carrier) for haemophilia may transmit the disease to sons. The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic (unviable in the later stage of life). The family pedigree of Queen Victoria shows a number of haemophilic descendants as she was a carrier of the disease.

MENDELIAN DISORDERS

(1) SICKLE-CELL ANAEMIA:

- This is an **autosome linked recessive trait** that can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous).
- The disease is controlled by a single pair of allele, **Hb^A and Hb^S.**
- Out of the three possible genotypes only homozygous individuals for Hb^S (Hb^SHb^s) show the diseased phenotype. Heterozygous (Hb^AHb^S) individuals appear apparently unaffected but they are carrier of the disssease as there is 50 per cent probability of transmission of the mutant gene to the progeny, thus exhibiting sickle-cell trait.
- The defect is caused by the **substitution of Glutamic acid (Glu) by Valine (Val)** at the sixth position of the beta globin chain of the haemoglobin molecule.
- The substitution of amino acid in the globin protein results due to the single base substitution **at the sixth codon of the beta globin gene from GAG to GUG.**

The mutant **haemoglobin molecule undergoes polymerisation under low oxygen tension** causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure.



(2) PHENYLKETONURIA:

- This inborn error of metabolism is also inherited as the **autosomal recessive trait.**
- The affected individual lacks an enzyme that converts the amino acid **phenylalanine into tyrosine.**
- As a result of this phenylalanine is accumulated and converted into phenylpyruvic acid and other derivatives. Accumulation of these in brain results in **mental retardation.** These are also excreted through urine because of its poor absorption by kidney.

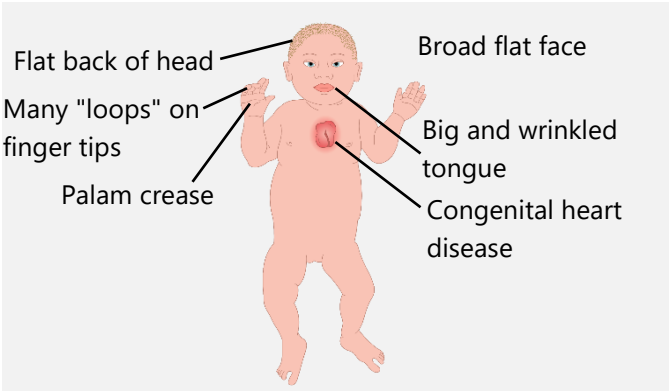
(3) THALASSEMIA :

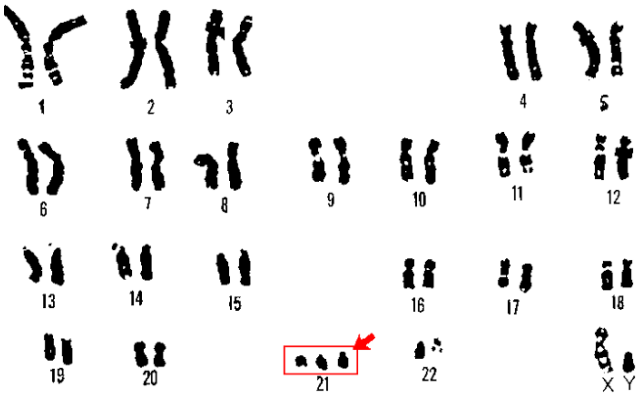
- This is also an **autosome-linked recessive blood disease** transmitted from parents to the offspring when both the partners are unaffected carrier for the gene (or heterozygous).
- The defect could be due to either mutation or deletion which ultimately results in reduced rate of synthesis of one of the globin chains (α and β chains) that make up haemoglobin.
- This causes the formation of abnormal haemoglobin molecules resulting into anaemia which is characteristic of the disease.
- Thalassemia can be classified according to which chain of the haemoglobin molecule is affected. In **α Thalassemia,** production of α globin chain is affected while in **β Thalassemia,** production of β globin chain is affected.
- α Thalassemia is controlled by two closely linked genes **HBA1 and HBA2onchromosome 16** of each parent and it is observed due to mutation or deletion of one or more of the four genes. The more genes affected, the less alpha globin molecules produced.
- While **β Thalassemia is controlled by a single gene HBB on chromosome 11** of each parent and occurs due to mutation of one or both the genes.

  Thalassemia differs from sickle-cell anaemia in that the former is a **quantitative** problem of synthesising too few globin molecules while the latter is a **qualitative problem** of synthesising an incorrectly functioning globin.

CHROMOSOMAL DISORDERS

- The chromosomal disorders on the other hand are caused due to absence or excess or abnormal arrangement of one or more chromosomes.
- Failure of segregation of chromatids during cell division cycle results in the gain or loss of a chromosome(s), **called aneuploidy.**
- For example, **Down's syndrome** results in the gain of extra copy of **chromosome 21.** Similarly, **Turner's syndrome** results due to **loss of an X chromosome** in human females.
- **Failure of cytokinesis after telophase** stage of cell division results in an increase in a whole set of chromosomes in an organism and, this phenomenon is known as **polyploidy.** This condition is often seen in plants. The total number of chromosomes in a normal human cell is 46 (23 pairs). Out of these 22 pairs are autosomes and one pair of chromosomes are sex chromosome. Sometimes, though rarely, either an additional copy of a chromosome may be included in an individual or an individual may lack one of any one pair of chromosomes. These situations are known as trisomy or monosomy of a chromosome, respectively. Such a situation leads to very serious consequences in the individual. Down's syndrome, Turner's syndrome, Klinefelter's syndrome are common examples of chromosomal disorders.
- **Down's Syndrome:** The cause of this genetic disorder is the presence of an additional copy of the chromosome number 21 (trisomy of 21). This disorder was first described by Langdon Down (1866). The affected individual is short statured with small round head, furrowed tongue and partially open mouth. Palm is broad with characteristic palm crease. Physical, psychomotor and mental development is retarded.





A representative figure showing an individual inflicted with Down's syndrome and the corresponding chromosomes of the individual.

- **Klinefelter's Syndrome:** This genetic disorder is also caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of **47, XXY**. Such an individual has overall masculine development, however, the feminine development (development of breast, i.e., Gynaecomastia) is also expressed. Such individuals are sterile.

- **Turner's Syndrome:** Such a disorder is caused due to the absence of one of the X chromosomes, i.e., **45 with X0**, Such females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters.

Question-1: What is the inheritance of colour blindness of both parents having a normal vision but mother has a recessive gene for colour blindness :-

	Son	Daughter
(1)	50%	Nil
(2)	100%	Nil
(3)	Nil	100%
(4)	Nil	Nil

Answer: (1) Son 50%, Daughter Nil

Question-2: What would be the nature of children if a colour blind woman marries a normal man :-

(1) Colour-blind daughter & normal sons

(2) Colour-blind sons and carrier daughters

(3) Normal sons & carrier daughters

(4) Normal sons & Normal daughters

Answer: (2) Colour-blind sons and carrier daughters

Explanation:

Use checker board method. Genotype of normal man and colour blind woman respectively: XY , X^cX^c

Question-3: Rarely females experience the physiological defect of haemophilia as they do so only when they are :-

(1) Carrier for the defect

(2) Wives of haemophilic husbands

(3) Homozygous for the defect

(4) Heterozygous for the defect

Answer: (3) Homozygous for the defect

Question-4: Sickle cell anaemia induces due to :-

(1) Change of Amino Acid in α – chain of Haemoglobin

(2) Change of Amino Acid in β – chain of Haemoglobin

(3) Change of Amino Acid in both α and β chain of Haemoglobin

(4) Change of Amino acid either α or β chain of Haemoglobin

Answer: (2) Change of Amino Acid in β – chain of Haemoglobin

Question-5: Sickel cell anemia is the result of _____mutation in the haemoglobin gene :-

(1) Frame shift

(2) Deletion

(3) Point

(4) None of the above

Answer: (3) Point

Question-6: In sickle cell anaemia :–

(1) The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of RBC

(2) Substitution of Glutamic acid by valine at the sixth position of the α-chain of haemoglobin

(3) The mutant haemoglobin undergoes polymerization under high oxygen tension causing the change in shape of RBC

(4) A-globin chain is modified

Answer: (1) The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of RBC

Question-7: Which of the following can be called a mutation :-

(1) The halfing of the chromosome number at meiosis

(2) The doubling of the chromosome after syngamy

(3) The possession of an additional chromosome

(4) All the above

Answer: (4) All the above

Question-8: Chromosome number 2n-1 is an example of :-

(1) Trisomy

(2) Euploidy

(3) Polyploidy

(4) Monosomy

Answer: (4) Monosomy

Question-9: The number of chromosomes a child with Down syndrome has is

(1) 45

(2) 46

(3) 47

(4) 48

Answer: (3) 47